ANDROGEN RECEPTOR GENE MUTATIONS DATABASE

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Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile: k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0001	PAIS	Substitut.	*	002 367	Glu⇒Lys GAA⇒AAA					high	20-50% reduction in mutant protein	Male	Ambiguous	pos	Choong et al; J Clin Invest. 98: 1423-1431, 1996
0960	CAIS	Substitut.		002 367	Glu⇒Lys GAA⇒AAA	27	13	low	high		Mother heterozygous carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0751	CAIS	Deletion		007 381	Leu⇒0 ΔCTG⇒						Elevated testosterone stop in codon 33 No WD development	Female	Normal	neg	Barbaro et al. Clin Endocrinol 66:822-826, 2007
0963	CAIS	Insertion		015 415	Pro⇒Ala CCG⇒	20	9				4nt insertion (GCCG) causing fs and stop in codon 82	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0624	CAIS	Deletion		039 477	Pro⇒0 ΔCCC⇒						In frame deletion of 3nt. removing proline				Jung et al. Human Genetics 114: 222, 2004
0836	CAIS	Deletion		040 480	Arg⇒0 ΔAGG⇒						1 nt del causing framshift & stop in codon 172	Female	Normal	pos	Decaestecker et al; Fertility & Sterility 89: 1260 e3-7, 2008
0910	Prostate cancer	Substitut.		043 490	Glu⇒Gly GAG⇒GGG						Treated with Flutamide -occurred in two cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0002	CAIS	Deletion		051	Gly⇒0 GGΔC⇒				zero		1 nt del causing frameshift & stop in Codon 180 1 aff sib	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0003	Prostate cancer	Substitut.		054 523	Leu⇒Ser TTG⇒TCG						Also Phe891Leu (TTT to CTT) mut. Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0004	Laryngeal cancer	Deletion		057	⇒ ⇒						30 nt. deletion Somatic mutation	Male	Normal		Urushibata et al; 10th. Int. Cong. Endocrinol Abstr. P3-706, 1996
0815	Testicular cancer	Deletion		057	Leu⇒0 CTG⇒						3 nt. deletion seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12:645 -655, 2005
0808	Liver cancer	Substitut.		057 532	Leu⇒Gln CTG⇒CAG						Somatic mutation	Male	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0005	Prostate cancer	Substitut.		057 532	Leu⇒Gln CTG⇒CAG						Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0786	MAIS	Insertion		057 531	⇒ ⇒						Leucine inserted - male inferility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0411	Mental Retard.	Deletion		058	⇒ ⇒	8		normal	normal		3 affected siblings - normal CAG = 23	Male	Normal	pos	Kooy et al; Am J Med Genet. 85: 389-393. 1999
0612	MAIS ?	Substitut.		058 535	Gln⇒Leu CAG⇒CTG						2 out of 62 patients with male infertility	Male	Normal		Lund et al; Fertlilty and Sterility 79(suppl 3): 1647-148, 2003
0897	CAIS	Insertion		058 535	Gln⇒ CAG⇒CCTG							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0006	Kennedy Syndrome	Insertion		058-078	⇒ ⇒	> 40					Expansion of polyglutamine repeat	Male	Normal		LaSpada et al; Nature 352:77, 1991
0007	Prostate cancer	Deletion		058-078	⇒ ⇒	18					Contraction of poly Gln repeats (24 to 18) Somatic mutation	Male	Normal		Schoenberg et al; Bioch. & Biophys Res Comm 198: 74-80 1994
0324	Prostate cancer	Deletion		058-078	⇒ ⇒	22					Deletion of 1polyGln repeat (23-22) Somatic mutation	Male	Normal		Watanabe et al; Jpn J Clin Oncol 27: 389 -393, 1997
0325	Prostate cancer	Insertion		058-078	⇒ ⇒	22					Insertion of 1polyGln repeat (21-22) in 2 diff patients.Som mut	Male	Normal		Watanabe et al; Jpn J Clin Oncol 27: 389 -393, 1997
0495	Prostate cancer	Deletion		058-078	⇒ ⇒	18					Contraction of poly Gln repeats (20 to 18) Somatic mutation	Male	Normal		Wallin et al; J Pathology 189: 559-653, 1999

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0692	CAIS	Substitut.		1 Nterm	059 540	Gln⇒Stop CAG⇒TAG			zero				Female	Normal		Holterhus et al; J Mol Med 2005
0008	CAIS	Substitut.	*	1 Nterm	060 540	Gln⇒Stop CAG⇒TAG			low	normal	high	Normal upregulation.	Female	Normal	neg	Zoppi et al; J Clin Inv 19:1105, 1993
0671	CAIS	Substitut.	*	1 Nterm	060 540	Gln⇒Stop CAG⇒TAG						bilateral inguinal hernia.	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0409	CAIS	Insertion or deletion		1 Nterm	060 540	Gln⇒Gln CAG⇒CAAG						either 1nt. insert or 2nt. del. -frameshift & stop in codon 80	Female	Normal		Zhu et al; J Clin Endocrinol & Metab 84: 1590-1594, 1999
0009	Prostate cancer	Substitut.		1 Nterm	064 550	Gln⇒Arg CAG⇒CGG						Also Leu830Pro (CTT to CCT) mut. Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0846	CAIS	Substitut.		1 Nterm	067 558	Gln⇒Stop CAG⇒TAG						Diag at 1Mo Bilateral gonadectomy at 18y	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0894	CAIS	Substitut.		1 Nterm	070 569	Gln⇒Stop CAG⇒TAG							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0881	MAIS ?	Substitut.		1 Nterm	070 570	Gln⇒Arg CAG⇒CGG	20,21,23					22 CAGs in blood azoospermia Sertoli Cell only Syndrome	Male	Normal		Hose et al; Fertility & Sterility 92: 390e9-e11, 2009
0787	PAIS?	Substitut.		1 Nterm	073 579	Gln⇒Stop CAG⇒TAG						Somatic mosiac 2/3 mutant to 1/3mutant	Female	Normal		Mueller et al. Hum Genet 119:681, 2006
0895	CAIS	Substitut.		1 Nterm	076 588	Gln⇒Stop CAG⇒TAG							Female	Normal	pos	Philibert et al; Fertility & Sterility 2009
0961	CAIS	Substitut.		1 Nterm	076 588	Gln⇒Stop CAG⇒TAG		18				Truncated CAG repeat ?	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0902	CAIS	Deletion		1 Nterm	076 589	Gln⇒ CAAG⇒CAC							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0807	Liver cancer	Substitut.		1 Nterm	078 593	Gln⇒Gln CAA⇒CAG						somatic mutation? present in tumor and non-tumor tissue	Male	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0962	CAIS	Insertion		1 Nterm	079 596	Glu⇒Arg GAG⇒	21	18				1nt insertion causing frameshift and stop in Codon 81	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0901	CAIS	Deletion		1 Nterm	080 600	Thr⇒ ΔACTAG⇒						5 base deletion	Female	Normal	pos	Philibert et al; Fertility & Sterility 2009
0965	CAIS	Deletion		1 Nterm	082	Pro⇒Ser CCC⇒	20	18	zero	zero		10 base deletion frameshift & stop in codon 169	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0964	CAIS	Substitut.		1 Nterm	084 612	Gln⇒Stop CAG⇒TAG	20		low	high			Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0416	CAIS	Insertion		1 Nterm	085 615	Gln⇒Gln CAG⇒CAAG	25		zero			1nt. insertion causing frameshift and stop in codon 91	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0796	CAIS	Substitut.		1 Nterm	086 618	Gln⇒Stop CAG⇒TAG			zero				Female	Normal		Jaaskelainen et al. Hum Mutat 27:291, 2006
0882	Prostate Cancer	Deletion		1 Nterm	086 618	Gln⇒0 ΔCAG⇒						+Q58L,T438P,G456S flutamide treatment	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0672	CAIS	Substitut.		1 Nterm	088 624	Gln⇒Stop CAG⇒TAG						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0806	Liver cancer	Substitut.		1 Nterm	089 629	Gln⇒His CAG⇒CAT						Somatic mutation	Male	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0529	CWR22R Prost. CA Cell line	Substitut.		1 Nterm	091 635	Glu⇒Asp ⇒	27	19				AR indep.+Leu57Gln & His 874Tyr mut. + Duplication of exon 3	Male	Normal		Chelnski et al. The Prostate 47: 66-75, 2001
0805	Liver cancer	Substitut.		1 Nterm	100 661	Arg⇒Lys AGA⇒AAA						somatic mutation ? present in tumor and non-tumor tissue	Male	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0417	CAIS	Deletion		1 Nterm	102 668	Pro⇒Pro CCΔC⇒CCG	12	25	zero			1 nt. deletion causing frameshift and stop in codon 172	Female	Normal	neg	Gottlieb et al; Hum Mutat. 14: 527-539, 1999

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0898	CAIS	Insertion		1 Nterm	107 682	Val⇒ GTC⇒GATC							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0010	Prostate cancer	Substitut.		1 Nterm	112 698	Gln⇒His CAG⇒CAT					AlsoTrp798Stop (TGG to TGA) mut. Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0418	CAIS	Substitut.		1 Nterm	113 699	Gln⇒Stop CAA⇒TAA	25	27				Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0802	CAIS	Substitut.		1 Nterm	118 714	Gln⇒Stop CAG⇒TAG					1 affected sister	Female	Normal	pos	Berg et al. J Pediatr 150:434-438, 2007
0631	CAIS	Substitut.		1 Nterm	119 718	Ser⇒Stop TCG⇒TAG	25				gonads located in inguinal canal. Same family as 0830	Female	Normal	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0830	CAIS	Substitut.		1 Nterm	119 718	Ser⇒Stop TCG⇒TAG					gonads located in inguinal canal. Same family as 0631	Female	Normal	pos	Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
0911	Prostate cancer	Substitut.		1 Nterm	119 719	Ser⇒Ser TCG⇒					Silent mutation Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442
0417	CAIS	Deletion		1 Nterm	125 738	Pro⇒Pro CCΔC⇒CCG	23	24	zero		1nt. deletion causing frameshift and stop at codon 172	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0011	CAIS	Deletion		1 Nterm	127 743	Arg⇒Arg AGAΔA⇒AGG			zero		1 nt deletion causing frameshift & stop in Codon 172	Female	Normal	neg	Batch et al; Hum Mol Genet 1: 497, 1992
0436	CAIS	Deletion		1 Nterm	127 743	Arg⇒Arg AGAΔA⇒AGG					1 nt deletion causing frameshift & stop in Codon 172	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0012	CAIS	Deletion		1 Nterm	140	⇒ ⇒					Deletion of Codons 140-148 Stop in Codon 154	Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0837	CAIS	Substitut.		1 Nterm	141 784	Lys⇒Stop AAG⇒ATG					Sertoli-Lydig tumor	Female	Normal		Jazabeck et al; Gynecol Endocrinol 23:499-504, 2007
0694	Prostate Cancer	Substitut.	*	1 Nterm	142 787	Gly⇒Val GGG⇒GTG	16		normal	normal	Increased response to DHT	Male	Normal		Chen et al; The Prostate 63:395-406, 2005
0516	CAIS	Substitut.		1 Nterm	153 819	Glu⇒Stop GAG⇒TAG						Female	Normal		Copelli et al; Asian J Androl 1: 73-77, 1999
0523	CAIS	Substitut.		1 Nterm	153 819	Glu⇒Stop GAG⇒TAG						Female	Normal		Gacobini et al. Hum Genet. 108; 176, 2001
0788	PAIS	Substitut.		1 Nterm	157 831	Ala⇒Thr GCT⇒ACT									Mueller et al. Hum Genet 119:681, 2006
0627	Prostate Cancer	Substitut.		1 Nterm	166 858	Gly⇒Ser GGC⇒AGC					Orch + bicalutamitide treatment Gleason 10 Somatic. mutation	Male	Normal		Haaplaa et al. Lab Invest. 81:1647-1651, 2001
0838	CAIS	Deletion		1 Nterm	166 858	Gly⇒0 ΔGGC⇒					Frameshift and stop in codon	Female	Normal		Jeske et al; J Pediatr Endocrinol Metab 20:893-908, 2007
0013	CAIS	Substitut.		1 Nterm	172 877	Leu⇒Stop TTA⇒TGA						Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0316	PAIS	Substitut.		1 Nterm	172 877	Leu⇒Stop TTA⇒TGA			low	normal	Somatic mosaic mut. causes partial virulization	Female	Ambiguous		Holterhus et al; J Clin Endocrinol. 82: 3584-3589, 1997
0420	CAIS	Substitut.		1 Nterm	172 877	Leu⇒Stop TTA⇒TGA	26	24	zero			Female	Normal	neg	Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0566	Prostate cancer	Substitut.		1 Nterm	176 190	Ser⇒Ser TCC⇒TCT					Som Mut. Exposed to Estrogen treat. Also Leu744Leu Silent	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0014	Prostate cancer	Substitut.		1 Nterm	180 901	Lys⇒Arg AAA⇒AGA					Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0912	Prostate cancer	Substitut.		1 Nterm	192 936	Leu⇒Phe CTT⇒TTT					Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442
0913	Prostate cancer	Substitut.		1 Nterm	192 937	Leu⇒Arg CTT⇒CGT					Treated with anti-androgens	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442

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0319	CAIS	Substitut.		194 943	Gln⇒Arg CAA⇒CGA						Also 1 nt deletion in Codon 597 causing a stop	Female	Normal		Komori et al; J Obstetrics & Gynocol. 23: 277-81, 1997
0551	Prostate cancer	Substitut.		198 955	Glu⇒Gly GAA⇒GGA						Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al; 37th meeting ASCO 20: Abstr, 1738 2001
0015	CAIS	Insertion		202	Glu⇒0 ⇒			zero			4 nt insertion causing frameshift & stop in Codon 232	Female	Normal	neg	Batch et al; Hum Mol Genet 1:497, 1992
0549	Prostate cancer	Substitut.		202 968	Glu⇒Glu GAA⇒GAG						Treated with bicalutamide - silent mutation- somat. mut.	Male	Normal		Taplin et al; 37th meeting ASCO 20: Abstr, 1738 2001
0395	Normal	Substitut.		205 977	Ser⇒Arg AGC⇒AGG						Homosexual individual	Male	Normal		Macke et al; Am J Human Genetics 53: 844-852, 1993
0437	CAIS	Deletion		208 985	Arg⇒Lys AAGA⇒AAG			zero			Frameshift & stop in codon 232 ?	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0376	MAIS	Substitut.		210 992	Arg⇒Arg AGG⇒AGA						Silent mutation	Male	Normal		Wang et al; Clinical Genetics 54: 185-192, 1998
0328	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mutation - polymorphism detected in 8% popul.	Male	Normal		Batch et al; Hum Mol Genet 1:497, 1992
0329	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mut.polymorph -detected in 14% of X chromosomes	Male	Normal		Hiort et al; Eur J Pediatr 153: 317-321, 1994
0330	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mutation polymorphism	Male	Normal		Lu et al; Clinical Genetics 49: 323-324. 1996
0377	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mutation polymorphism	Male	Normal		Wang et al; Clinical Genetics 54: 185-192, 1998
0396	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mut.polymorph detected in 10% of X chromosomes	Male	Normal		Macke et al; Am J Human Genetics 53: 844-852, 1993
0378	MAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mutation polymorphism - 4 patients with	Male	Normal		Wang et al; Clinical Genetics 54: 185-192, 1998
0421	CAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA	22	24	v low			Silent mutation - negligible level of mRNA & hAR	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0422	CAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA	21	23	normal	normal		Silent mutation -	Female	Normal	neg	Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0423	PAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA	23	24	v low			Silent mutation -	Male	Ambiguous		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0424	PAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA	19	24	normal	high		Silent mutation -	Male	Ambiguous	pos	Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0425	MAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA	20	16	normal	high		Silent mutation -	Male	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0705	MAIS	Substitut.		211 995	Glu⇒Glu GAG⇒GAA				normal			Male	Ambiguous		Deeb et al; Clinical Endocrinol 63:56-62, 2005
0701	Normal	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Silent mut.polymorph 65% Ivory Coast, 28% N African freqs.	Male	Normal		Estaban et al; Am J Hum Biol 17: 690-695, 2005
0756	Prostate cancer	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0757	Prostate cancer	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0758	Prostate cancer	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0914	Prostate cancer	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
1019	Premature ovarian failure	Substitut.		211 995	Glu⇒Glu GAG⇒GAA						Present in 17.85% of patients & 15% of controls	Female	Normal		Panda et al. Gyncol Endocrinol 2010

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0379	MAIS	Substitut.	1 Nterm	*	214 1005	Gly⇒ Arg GGG⇒ AGG	27	23	normal	normal	norm		severe oligospermia-20% lower transactivation	Male	Normal		Wang et al; Clinical Genetics 54: 185-192, 1998
0707	PAIS	Substitut.	1 Nterm		214 1005	Gly⇒ Arg GGG⇒ AGG				normal				Female	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0708	PAIS	Substitut.	1 Nterm		214 1005	Gly⇒ Arg GGG⇒ AGG				normal				Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0709	PAIS	Substitut.	1 Nterm		214 1005	Gly⇒ Arg GGG⇒ AGG				high				Female	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0380	Normal	Substitut.	1 Nterm		214 1005	Gly⇒ Arg GGG⇒ AGG								Male	Normal		Wang et al; Clinical Genetics 54: 185-192, 1998
0016	CAIS	Insertion	1 Nterm		215	Ala⇒ Gly GCT⇒ GGCT							1 nt insertion causing frameshift & stop in Codon 232	Female	Normal	neg	Hiort et al; Hum Mol Genet 3: 1163-1166 1994
0966	CAIS	Deletion	1 Nterm		219	Ser⇒ TCC⇒	18	17	zero	zero			I nt deletion causing frameshift & stop in Codon 224	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0695	Prostate cancer	Substitut.	1 Nterm	*	221 1023	Asp⇒ His GAC⇒ CAC	16		normal	normal			Increased response to DHT	Male	Normal	neg	Chen et al; The Prostate 63:395-406, 2005
0548	Prostate cancer	Substitut.	1 Nterm		222 1026	Asn⇒ Asp AAT⇒ GAT							Treated with flutamide also Thr877Ala - somatic	Male	Normal		Taplin et al; 37th meeting ASCO 20: Abstr, 1738 2001
0650	CAIS	Substitut.	1 Nterm		223 1031	Tyr⇒ Stop TAC⇒ TAG							Woolfian remnants	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0917	Prostate cancer	Substitut.	1 Nterm		227 1057	Thr⇒ Ala ACT⇒ GCT							Both treated and untreated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0915	Prostate cancer	Substitut.	1 Nterm		227 1057	Thr⇒ Cys ACT⇒ TGC							Untreated - Occurred in two cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0916	Prostate cancer	Substitut.	1 Nterm		227 1057	Thr⇒ Cys ACT⇒ TGT							Both treated and untreated - Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0885	Prostate cancer	Substitut.	1 Nterm		229 1057	Thr⇒ Cys ACT⇒ TGT							+Q58L, A251V, W433L, Flutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0706	PAIS	Substitut.	1 Nterm	*	232 1061	Ser⇒ Leu TCT⇒				high		*		Male	Normal		Giwercman et al. Clin Endocrinol 54: 827 -834, 2001
0531	MAIS	Substitut.	1 Nterm	*	233 1061	Asn⇒ Lys AAC⇒			normal			*	Azoospermia - transactivation 46% of wt	Male	Normal		Giwercman et al. Clin Endocrinol 54: 827 -834, 2001
0967	CAIS	Insertion	1 Nterm		239 1078	Lys⇒ Stop AAG⇒ ATAG	21	18						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0789	PAIS?	Substitut.	1 Nterm		246 1106	Gly⇒ Val GGC⇒ GTC											Mueller et al. Hum Genet 119:681, 2006
0918	Prostate Cancer	Substitut.	1 Nterm		251 1119	Ala⇒ Val GCG⇒ GTG							Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0883	Prostate Cancer	Substitut.	1 Nterm		253 1119	Glu⇒ Lys GAG⇒ AAG							+ L444S, R484C, K609E, R787X, L797P,L872P, +Flu	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0350	CAIS	Substitut.	1 Nterm	*	255 1126	Leu⇒ Pro CTG⇒ CCG						*	Also Gly820Ala mut. Extra mutation causes greater thermolability	Female	Normal		Tanaka et al; Gynocol Endocrinol 12: 75-82, 1998
0845	CAIS?	Insertion	1 Nterm		263 1145	Gly⇒ 0 GGG⇒ GgGG											Mueller; Hum Genet 123:105, 2008
0017	Prostate cancer	Substitut.	1 Nterm		266 1159	Met⇒ Thr ATG⇒ ACG							Also Leu574Pro (CTG to CCC) mut. Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0018	Prostate cancer	Substitut.	1 Nterm		269 1167	Pro⇒ Ser CCA⇒ TCA							Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0780	MAIS	Substitut.	1 Nterm		270 1170	Leu⇒ Phe CTT⇒ TTT							Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006

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0019	CAIS	Deletion	1 Nterm		272 1178	Gly⇒Gly GGΔA⇒GGG			zero			1 nt deletion causing frameshift & stop in Codon 301	Female	Normal		Bruggenwirth et al; J Steroid Biochem Mol Biol 58: 569-575,1996
0606	CAIS	Substitut.	1 Nterm		287 1221	Glu⇒Stop GAA⇒TAA			low	normal		low expression of WT AR - Somatic mosaicism	Female	Normal		Holterhaus et al; Genome Biology 4: R37
0556	Prostate cancer	Substitut.	1 Nterm		296 1250	Ser⇒Arg AGC⇒AGA						Poor differentiation of CaP. Germline mutation ?	Male	Normal		Yu et al; Sheng Wu Hua Xue 32: 459-462, 2000
0874	Prostate cancer	Substitut.	1 Nterm		296 1250	Ser⇒Arg AGC⇒AGA			normal			Transcrip activity activarted by estradiol and progesterone	Male	Normal		Li et al; Clin Exp Pharmacolo Physiol 35: 1252-1257, 2008
0816	Testicular cancer	Substitut.	1 Nterm		297 1251	Ala⇒Thr GCA⇒ACA						Seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12:645 -655, 2005
0550	Prostate cancer	Substitut.	1 Nterm		334 1359	Ser⇒Pro TCC⇒CCC						Treated with flutamide somatic mutation	Male	Normal		Taplin et al; 37th meeting ASCO 20: Abstr, 1738 2001
0398	Prostate cancer	Substitut.	1 Nterm		340 1381	Pro⇒Leu CCG⇒CTG						Somatic mutation Stage 3 tumor	Male	Normal		Castagnaro et al; Verh. Dtsch. Ges. Path. 77: 119-123, 1993
0779	MAIS	Substitut.	1 Nterm		340 1381	Pro⇒Leu CCG⇒CTG						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0968	CAIS	Deletion	1 Nterm		346 1397	Tyr⇒Gln ΔTAΔC⇒CAA	21	17				2 nt deletion causing fs & stop in Codon 499	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0903	CAIS	Deletion	1 Nterm		347 1401	Lys⇒Ile AΔAG⇒ATC							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0903	CAIS	Deletion	1 Nterm		347 1407	Gly⇒ ⇒						22 bp deletion	Female	Normal	pos	Philibert et al; Fertility & Sterility 2009
0020	CAIS	Substitut.	1 Nterm		353 1419	Glu⇒Stop GAG⇒TAG	21	23	low			low specific binding with MB only-mRNA < 20%	Female	Normal	neg	Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0781	MAIS	Substitut.	1 Nterm		353 1419	Glu⇒Gln GAG⇒CAG						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0919	Prostate Cancer	Substitut.	1 Nterm		357 1431	Ala⇒Thr GCT⇒ACT						Bicalutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0920	Prostate Cancer	Substitut.	1 Nterm		357 1432	Ala⇒Val GCT⇒GTT						Bicalutamide treated Occurred in two cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0921	Prostate cancer	Substitut.	1 Nterm		360 1441	Arg⇒His CGC⇒CAC						Both treated and untreated Occurred in two cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0021	CAIS	Substitut.	1 Nterm		371 1474	Gly⇒Stop GGA⇒TGA						Somatic instability of polyglycine tract	Female	Normal	pos	Davies et al; Clinical Endocrinology 43: 69 -77, 1995
1003	PAIS	Substitut.	1 Nterm		378 1596	Pro⇒Arg CCT⇒CGT	17	18				Prenatal diagnosis	Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0899	CAIS	Insertion	1 Nterm		381 1505	Pro⇒ ⇒						201 bp insertion-dup (CCTCCGCCGCC)	Female	Normal	pos	Philibert et al; Fertility & Sterility 2009
0908	CAIS	Substitut.	1 Nterm		390 1530	Pro⇒Ser CCG⇒TCG			zero				Female	Normal		Appari et al; J Molecular Med: 87:623 -632.,2009
0950	PAIS	Substitut.	1 Nterm		390 1530	Pro⇒Ser CCG⇒TCG						Micropenis only	Male	Ambiguous		Bhangoo et al. Asian J Androl 12:561-566. 2010
1004	PAIS	Substitut.	1 Nterm		390 1530	Pro⇒Ser CCG⇒TCG	22	19	normal	normal			Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0338	MAIS	Substitut.	1 Nterm	*	390 1530	Pro⇒Ser CCG⇒TCG						Oligospermia	Male	Normal		Hiort et al; J Clin Endocrinol & Metab 85: 2810-2815, 2000
0504	MAIS	Substitut.	1 Nterm	*	390 1530	Pro⇒Ser CCG⇒TCG						Oligospermia	Male	Normal		Hiort et al; J Clin Endocrinol & Metab 85: 2810-2815, 2000
0767	MAIS	Substitut.	1 Nterm		390 1530	Pro⇒Ser CCG⇒TCG						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006

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0817	Testicular cancer	Substitut.	1 Nterm		390 1530	Pro⇒Ser CCG⇒TCG						Seminoma	Male	Normal		Garolla et al. Encdorine Related Cancer 12:645 -655, 2005
0608	Prostate cancer	Substitut.	1 Nterm		390 1531	Pro⇒Leu CCG⇒TCG			zero				Female	Normal		Holterhaus et al; Genome Biology 4: R37
0547	Prostate cancer	Substitut.	1 Nterm		390 1531	Pro⇒Leu CCG⇒CTG						Treated with flutamide also Asn756Asp -	Male	Normal		Taplin et al; 37th meeting ASCO 20: Abstr, 1738, 2001
0022	CAIS	Substitut.	1 Nterm		390 1531	Pro⇒Arg CCG⇒CGG	20	24	zero			+ substs. Glu211Glu GAGtoGAA&Gln443 Arg(CAGtoCGG)	Female	Normal	pos	Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0790	CAIS	Substitut.	1 Nterm		393 1531	Tyr⇒Stop TAC⇒TAG							Female	Normal		Mueller et al. Hum Genet 119:680, 2006
0896	CAIS	Substitut.	1 Nterm		393 1531	Tyr⇒Stop TAC⇒TAG							Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0426	CAIS	Substitut.	1 Nterm		403 1569	Gln⇒Stop CAG⇒TAG	28	23	zero			mRNA < 20%	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0689	PAIS	Deletion	1 Nterm	*	409 1587	Leu⇒0 GTG⇒						Deletion of codons 409-411. Normal twin has same deletion	Male	Ambiguous	pos	Holterhus et al; Exp Clin Endocrinol diabetes 113: 457-463,
0690	PAIS	Substitut.	1 Nterm	*	411 1594	Ser⇒Asn AGC⇒AAC						Patient responded to testosterone treat at 12yrs.	Male	Ambiguous		Holterhus et al; Exp Clin Endocrinol diabetes 113: 457-463,
0922	Prostate cancer	Substitut.	1 Nterm		414 1602	Gly⇒Ser GGC⇒AGC						Bicalutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0923	Prostate cancer	Substitut.	1 Nterm		414 1603	Gly⇒Asp GGC⇒GAC						Bicalutamide treated occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0691	PAIS	Substitut.	1 Nterm	*	432 1657	Ser⇒Phe TCC⇒TTC							Male	Ambiguous		Holterhus et al; Exp Clin Endocrinol diabetes 113: 457-463,
0889	Prostate Cancer	Substitut.	1 Nterm		433 1660	Trp⇒Leu TGG⇒TTG						+ΔQ86,T227C, T438P,T497I,V508L Q867X,Bical treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0924	Prostate Cancer	Substitut.	1 Nterm		433 1660	Trp⇒Leu TGG⇒TTG							Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0925	Prostate Cancer	Substitut.	1 Nterm		433 1661	Trp⇒Cys TGG⇒TGC						Both treated and untreated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0926	Prostate Cancer	Substitut.	1 Nterm		438 1674	Thr⇒Pro ACA⇒CCA						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0890	Prostate Cancer	Substitut.	1 Nterm		438 1675	Thr⇒Ile ACA⇒ATA						+Q58L, ΔQ86, G457D	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0759	Prostate cancer	Substitut.	1 Nterm		449 1708	Gly⇒Asp GGT⇒GAT						patient lower Gleason score than patient -wt AR- som mutation				Sanchez et al. BJU Int 98:1320-1325, 2006
0760	Prostate cancer	Substitut.	1 Nterm		451 1714	Gly⇒Asp GGT⇒GAT						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0969	CAIS	Substitut.	1 Nterm		453 1719	Gly⇒Ser GGT⇒AGT	22	17				Also Y571C mutation 2 affected aunts	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0888	Prostate Cancer	Substitut.	1 Nterm		454 1722	Gly⇒Ser GGC⇒AGC						+Q58L, ΔQ86, A251V, Bicalutmatide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0927	Prostate Cancer	Substitut.	1 Nterm		454 1722	Gly⇒Ser GGC⇒AGC						Treated with anti-androgens Occurred in more than 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0884	Prostate Cancer	Substitut.	1 Nterm		455 1726	Gly⇒Asp GGC⇒GAC						+ΔQ86, V716M, Flutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0928	Prostate Cancer	Substitut.	1 Nterm		455 1726	Gly⇒Asp GGC⇒GAC						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0438	CAIS	Deletion	1 Nterm		461 1745	Gly⇒Gly GGΔC⇒GGG			zero			1 nt. deletion causing frameshift	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000

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0410	CAIS	Deletion	1 Nterm	472 1776	Glu⇒Gly GΔAG⇒GGC	24	22				2nt. del causing frameshift & stop in cod 499- mRNA 50%	Female	Normal		Thiele et al; J Clin Endocrinol & Metab 84: 1751-1753. 1999
0693	CAIS	Deletion	1 Nterm	472 1776	Gly⇒Gly GΔAG⇒GGC	21	28	zero			2nt. del causing frameshift & stop in cod 499- mRNA 50%	Female	Normal		Holterhus et al; J Mol Med 2005
0427	CAIS	Deletion	1 Nterm	473 1779	Glu⇒Gly GΔAG⇒GGC	26	26	zero			2nt. del causing frameshift & stop in cod 499 -mRNA 50%	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0782	MAIS	Substitut.	1 Nterm	* 474 1783	Ala⇒Val GCG⇒GTG						male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0821	MAIS	Substitut.	1 Nterm	* 474 1783	Ala⇒Val GCG⇒GTG	14	17				male infertility, low T severe hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0822	MAIS	Substitut.	1 Nterm	* 474 1783	Ala⇒Val GCG⇒GTG	24	17				male infertility, normal T	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0823	MAIS	Substitut.	1 Nterm	* 474 1783	Ala⇒Val GCG⇒GTG	19	17				male infertility, normal T, left cryptorchidism	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0804	Liver cancer	Substitut.	1 Nterm	480 1802	Tyr⇒Tyr TAC⇒TAT						Somatic mutation	Female	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0024	CAIS	Substitut.	1 Nterm	480 1802	Tyr⇒Stop TAC⇒TAA	15	15	zero			Normal 110KD AR produced at 25% of normal level	Female	Normal		Gottlieb et al; Hum Mutat. 14: 527-539, 1999
0651	CAIS	Substitut.	1 Nterm	480 1802	Tyr⇒Stop TAC⇒TAA						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0929	Prostate cancer	Substitut.	1 Nterm	484 1811	Arg⇒Cys CGG⇒						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0930	Prostate cancer	Substitut.	1 Nterm	484 1811	Arg⇒Thr CGG⇒AGG						Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0546	CAIS	Deletion	1 Nterm	487 1821	Gln⇒Stop CAG⇒TAG						3 affected siblings Variable phenotype	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0439	CAIS	Deletion	1 Nterm	488 1824	Gly⇒0 ⇒			low			5 nt. deletion causing frameshift	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0440	CAIS	Substitut.	1 Nterm	491 1833	Gly⇒Ser GGC⇒AGC			low	low			Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0891	Prostate Cancer	Substitut.	1 Nterm	497 1852	Thr⇒Ile ACC⇒ATC						Also R484C Bicalutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0931	Prostate Cancer	Substitut.	1 Nterm	497 1852	Thr⇒Ile ACC⇒ATC						Both treated and not treated Occured in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0932	Prostate Cancer	Substitut.	1 Nterm	498 1854	Ala⇒Thr GCA⇒ACA						Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0933	Prostate Cancer	Substitut.	1 Nterm	498	Ala⇒Val GCA⇒GTA						Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0936	Prostate Cancer	Substitut.	1 Nterm	499 1859	Pro⇒Pro CCT⇒						Treated with ant-androgens Occurred in more than 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0905	CAIS	Indel	1 Nterm	502 1866	Trp⇒ TGG⇒						Indel between c1866 -1871	Female	Normal	neg	Philibert et al; Fertility & Sterility 2009
0025	CAIS	Substitut.	1 Nterm	502 1867	Trp⇒Stop TGG⇒TAG							Female	Normal	pos	Bruggenwirth et al; J Steroid Biochem Mol Biol 58: 569-575, 1996
0783	MAIS	Substitut.	1 Nterm	506 1879	Gly⇒Asp GGC⇒GAC						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0886	Prostate Cancer	Substitut.	1 Nterm	508 1884	Val⇒Leu GTG⇒CTG						Bicalutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0934	Prostate Cancer	Substitut.	1 Nterm	508 1884	Val⇒Leu GTG⇒CTG						Flutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009

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0935	Prostate Cancer	Substitut.	1 Nterm	508 1885	Val⇒Gly GTG⇒GGG						Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0339	MAIS	Substitut.	1 Nterm	* 511 1895	Val⇒Val GTG⇒GTA						Oligospermia caused by silent mutation	Male	Normal		Hiort et al; 80th US Endo Soc Meeting, Abstr P2-38, 1998
0876	CAIS	Substitut.	1 Nterm	513 1902	Tyr⇒Stop TAT⇒TAA							Female	Normal	pos	Radpour et al; J of Andrology 30:230-232, 2009
0567	Prostate cancer	Substitut.	1 Nterm	514 1903	Pro⇒Ser CCC⇒TCC						Somatic mut. Orchiectomy + EMP Horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0686	Normal	Substitut.	1 Nterm	516 1907	Pro⇒Ser CCC⇒AGC						Patient suffered from phobia		Normal		Yan et al; Psychiatric Genet 14: 57-60, 2004
0568	Prostate cancer	Substitut.	1 Nterm	524 1932	Gly⇒Ser GGC⇒AGC						Somatic mut. Orchi + EMP treat. Also Trp526stop mutation	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0569	Prostate cancer	Substitut.	1 Nterm	524 1933	Gly⇒Asp GGC⇒GAC						Somatic mut. Orchiectomy + EMP Horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0026	Prostate cancer	Substitut.	1 Nterm	528 1945	Asp⇒Gly GAT⇒GGT						Somatic mutation	Male	Normal		Tilley et al; Chemical Cancer Res. 2: 277-285, 1996
900	CAIS	Insertion	1 Nterm	530 1951	Tyr⇒ ⇒						GCTA insertion	Female	Normal		Philibert et al; Fertility & Sterility 2009
0570	Prostate cancer	Substitut.	1 Nterm	533 1959	Pro⇒Ser CCT⇒TCT						Somatic mut. Orchiectomy + EMP horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0027	CAIS	Substitut.	1 Nterm	534 1964	Tyr⇒Stop TAC⇒TAG			zero				Female	Normal	neg	McPhaul et al; FASEB J 5:2910-15, 1991
0028	CAIS and mental retardation	Deletion	1-8		⇒ ⇒			zero			Termini not yet defined	Female	Normal	neg	Trifiro et al; Mol Cell Endocrinol 75:37-47, 1991
0029	CAIS	Deletion	1-8		⇒ ⇒			zero				Female	Normal	pos	Quigley et al; J Clin Endocrinol Metab 74:927, 1992
0030	CAIS	Deletion	1-8		⇒ ⇒			zero				Female	Normal	pos	Hiort et al; Am J Med Genet. 63: 218-22, 1996
0435	CAIS	Deletion	1-8		⇒ ⇒			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0031	CAIS	Deletion	2		⇒ ⇒							Female	Normal		Quigley et al; J Cell Biochem Suppl 16C; Abstr L323, 1992
0579	CAIS	Deletion	2		⇒ ⇒						No immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0580	CAIS	Deletion	2		⇒ ⇒						No immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0441	CAIS	Duplicat.	2		⇒ ⇒						Duplication of exon 2	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0652	CAIS	Duplicat.	2		⇒ ⇒						Duplication of exon 2 sister of 653	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0653	CAIS	Duplicat.	2		⇒ ⇒						Duplication of exon 2 sister of 652	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0970	CAIS	Deletion	2 DBD	545 1996	His⇒ CAT⇒	23	17				17bp del. at 2/3 splice site resulting in fs & stop in Codon 547?	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0032	PAIS	Substitut.	2	547 2003	Leu⇒Phe TTG⇒TTT			low	high		Also has Trp741Cys (TGG to TGT) mutation	Male	Ambiguous	pos	Karl et al; 76th US Endo Soc Meeting, Abstr 1735, 1994
0673	PAIS	Substitut.	2	547 2003	Leu⇒Phe TTG⇒TTT								Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0768	MAIS	Substitut.	2	547 2003	Leu⇒Phe TTG⇒TTT						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006

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0357 Prostate cancer	Deletion	2		547 2003	Leu⇒Leu TTΔG⇒TTC						Frameshift - somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0824 MAIS	Substitut.	2	*	547 2003	Leu⇒Phe TTG⇒TTT	19	17				Azoospermia, Normal T, Left varicocele, Sertoli 0nly syndrom	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0825 MAIS	Substitut.	2	*	547 2003	Leu⇒Phe TTG⇒TTT	19	18				Male infertility, low T	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0033 MAIS	Substitut.	2		548 2004	Pro⇒Ser CCC⇒TCC						Distal hypospadia, variable penetrance in family members	Male	Near-normal male	pos	Sutherland et al; J of Urology 156: 828-831, 1996
0023 CAIS		2		 2001	⇒ ⇒						Duplication of 8nt. # 2011-2018 frameshift & stop in Codon 563	Female	Normal		Lumbroso et al; 10th Int Cong of Endocrinol, Abstr P1-182, 1996
0358 Prostate cancer	Deletion	2		554 2023	Pro⇒Pro CCΔA⇒CCC						Frameshift - somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0359 Prostate cancer	Deletion	2		554 2023	Pro⇒Pro CCΔA⇒CCC						Frameshift - somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0952 Prostate cancer	Substitut.	2 DBD		558 2034	Thr⇒Ser ACC⇒TCC						Germline mutation early onset disease in African-American	Male	Normal	pos	Hu et al. Asian J Androl 12:336-343, 2010
0034 CAIS	Substitut.	2 DBD	*	559 2038	Cys⇒Tyr TGC⇒TAC			normal	normal			Female	Normal	neg	Zoppi et al; Mol Endocrinol 6:409, 1992
0840 CAIS	Insertion	2 DBD		562 2045	Ile⇒ ATCins CTG ⇒						Insertion of CTG	Female	Normal		Jeske et al; J Pediatr Endocrinol Metab 20:893-908, 2007
0797 CAIS	Substitut.	2 DBD		562 2047	Cys⇒Tyr TGT⇒TAT							Female	Normal		Jaaskelainen et al. Hum Mutat 27:291, 2006
0035 PAIS	Substitut.	2 DBD		568 2064	Gly⇒Trp GGG⇒TGG			normal	normal			Female	Normal		Lobaccaro et al; Clin Endocrinol, 40:297, 1994
0674 PAIS	Substitut.	2 DBD		568 2064	Gly⇒Trp GGG⇒TGG										Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0037 PAIS	Substitut.	2 DBD		568 2065	Gly⇒Val GGG⇒GTG			normal	normal						Chang et al; 73rd US Endo Soc Meeting, Abstr 28, 1991
0036 PAIS	Substitut.	2 DBD		568 2065	Gly⇒Val GGG⇒GTG			normal			Severe hypospadia	Male	Ambiguous		Allera et al: J Clin Endocrinol & Metab 80: 2697-2699, 1995
0971 CAIS	Substitut.	2 DBD		568 2065	Gly⇒Glu GGG⇒GAG	26	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0545 PAIS	Substitut.	2 DBD		571 2073	Tyr⇒His TAT⇒CAT	21					sibling of 0747	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0558 PAIS	Substitut.	2 DBD		571 2073	Tyr⇒His TAT⇒CAT						DHT therapy effective	Male	Ambiguous		Foresta et al; Am J Med Genet 107: 259-260, 2002
0747 PAIS	Substitut.	2 DBD		571 2073	Tyr⇒His TAT⇒CAT						sibling of 0545	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0748 PAIS	Substitut.	2 DBD		571 2073	Tyr⇒His TAT⇒CAT			normal	low		nephew of 0545 & 0747	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0769 MAIS	Substitut.	2 DBD		571 2073	Tyr⇒His TAT⇒CAT						male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0826 PAIS?	Substitut.	2 DBD	*	571 2073	Tyr⇒His TAT⇒CAT	19	19				male infertility	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0032 CAIS	Substitut.	2 DBD	*	571 2974	Tyr⇒Cys TAT⇒TGT							Female	Normal		Komori et al; Arch Gynecol & Obstetrics 261: 95-100, 1998
0972 CAIS	Substitut.	2 DBD		573 2079	Ala⇒Asp GCT⇒ACT	24	18				1 affected sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0038 CAIS	Substitut.	2 DBD		573 2080	Ala⇒Asp GCT⇒GAT			normal			Defective DNA binding & transactivation	Female	Normal	neg	Bruggenwirth et al; J Steroid Biochem Mol Biol 58: 569-575, 1996

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0489 Prostate Cancer	Substitut.	2 DBD		575 2085	Thr⇒Ala ACA⇒GCA						Somatic Mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0810 Prostate Cancer	Substitut.	2 DBD	*	575 2085	Thr⇒Ala ACA⇒GCA						Som Mut. also T877A T575 bindisto AR- non-specific	Male	Normal		Monge et al.Cell Mol Life Sci 63:487-497, 2006
0039 CAIS	Substitut.	2 DBD	*	576 2088	Cys⇒Arg TGT⇒CGT			normal	normal			Female	Normal	pos	Zoppi et al; Mol Endocrinol 6:409, 1992
0040 CAIS	Substitut.	2 DBD		576 2089	Cys⇒Phe TGT⇒TTT			normal	normal			Female	Normal		Chang et al; 73rd US Endo Soc Meeting, Abstr 28, 1991
0407 CAIS	Substitut.	2 DBD		576 2089	Cys⇒Phe TGT⇒TTT						Lack of DNA binding -19 members of same family 3 testis tumors	Female	Normal	pos	Hooper et al; Cinical Genetics 65: 183-190, 2004
0554 PAIS	Substitut.	2 DBD	*	577 2091	Gly⇒Arg GGA⇒AGA			normal	normal	high	Alters affinity & selectivity of AR-ARE interactions				Nguyen et al; Mol Endocrinol 15:1790 -1802, 2001
0509 PAIS	Substitut.	2 DBD	*	578 2095	Ser⇒Thr AGC⇒ACC			normal			partial tranactivation in COS cells	Male	Ambiguous		Giwercman et al; Hormone Research 53: 83-88, 2000
0043 CAIS	Deletion	2 DBD		579 2099	Cys⇒Cys TGΔC⇒TGA			zero			Single nt. deletion causing frameshift & stop in Codon 619	Female	Normal		Imai et al, Annals of Clin Biochem, 32: 482 -486, 1995
0041 CAIS	Substitut.	2 DBD		579 2098	Cys⇒Tyr TGC⇒TAC							Female	Normal		Sultan et al, J Steroid Biochem & Mol Biol:46
0042 CAIS	Substitut.	2 DBD	*	579 2098	Cys⇒Phe TGC⇒TTC			normal	normal		Reduced transcription & DNA binding	Female	Normal	pos	Imasaki et al; Mol & Cell Endocrinol 120: 15 -24, 1996
0973 CAIS	Substitut.	2 DBD	*	579 2099	Cys⇒Trp TGC⇒TGG	23	17	low	low		Mother & 1 aunt heterozygous carriers, 1 aunt affected	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0487 Prostate Cancer	Substitut.	2 DBD		580 2101	Lys⇒Arg AAG⇒AGG						Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0843 PAIS	Substitut.	2 DBD		580 2101	Lys⇒Arg AAG⇒AGG						Attempted sex reassignment at 31 did not succeed	Female	Ambiguous		Katsumata et al; Endocr J 55:225-228, 2008
0044 CAIS	Substitut.	2 DBD	*	581 2103	Val⇒Phe GTC⇒TTC			normal	normal			Female	Normal		Lumbroso et al; Fertil Steril, 60:814, 1993
0675 CAIS	Substitut.	2 DBD	*	581 2103	Val⇒Phe GTC⇒TTC						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0676 CAIS	Substitut.	2 DBD	*	581 2103	Val⇒Phe GTC⇒TTC						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0719 PAIS	Substitut.	2 DBD		581 2103	Val⇒Leu GTC⇒CTC						neice also affetcted	Female	Ambiguous	pos	Ledig et al: Horm Res 63:263-269, 2005
0045 CAIS	Deletion	2 DBD	*	582 2104-6	Phe⇒0 GATCTT⇒GTC	22	23	low	normal		3 nt. del - Phe 582 del 2nt. from 581,1nt. 582. 581 still Val	Female	Normal	neg	Beitel et al; Hum Mol Genet, 3:21, 1994
0442 CAIS	Deletion	2 DBD		582 2106-8	Phe⇒0 TTC⇒			normal	normal		3 nt. del - of Phe	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0597 CAIS	Deletion	2 DBD		582 2106-8	Phe⇒0 TTC⇒						3 nt. del - of Phe	Female	Normal		Scheiber et al; J Pedatric Endocrinol & Metab. 16: 367-373,
0047 PAIS	Substitut.	2 DBD		582 2107	Phe⇒Ser TTC⇒TCC			zero				Female	Ambiguous		Hiort et al; Hum Mol Genet 3: 1163-1166 1994
0046 PAIS	Substitut.	2 DBD		582 2107	Phe⇒Tyr TTC⇒TAC			normal	normal		Reduced transcription & DNA binding	Female	Ambiguous	pos	Imasaki et al; Mol & Cell Endocrinol 120: 15 -24, 1996
0974 CAIS	Substitut.	2 DBD		583 2110	Phe⇒Leu TTC⇒TTG	29	17				Mother & 2 sisters heterozygous carriers 1 sister & 1 aunt affct	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0048 CAIS	Substitut.	2 DBD		585 2116	Arg⇒Lys AGA⇒AAA							Female	Normal		Sultan et al; J Steroid Biochem & Mol Biol:46
0049 CAIS	Deletion	2-8			⇒ ⇒			zero			Similar 2-8 deletion in 2 different families	Female	Normal		Jakubiczka et al; Human Mutation 9: 57 -61, 1997

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0050	CAIS	Deletion	3 DBD	*		⇒ ⇒			high	normal		Produces internally deleted protein	Female	Normal	pos	Quigley et al; Mol Endocrinol 6:1103, 1992
0051	CAIS	Deletion	3 DBD			⇒ ⇒							Female	Normal	pos	Hiort et al; Am J Med Genet. 63: 218-22, 1996
0443	CAIS	Deletion	3 DBD			⇒ ⇒			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0444	CAIS	Deletion	3 DBD			⇒ ⇒							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0488	Prostate Cancer	Substitut.	3 DBD		586 2119	Ala⇒ Val GCC⇒ GTC						Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0877	CAIS	Substitut.	3 DBD	*	586 2119	Ala⇒ Asp GCC⇒ GAC			normal			Zero transactivation activity	Female	Normal	pos	Rajender et al; Fert Steril 91:933.e23-28, 2009
0490	Prostate Cancer	Substitut.	3 DBD		587 2121	Ala⇒ Ser GCT⇒ TCT						Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0052	CAIS	Substitut.	3 DBD	*	590 2130	Lys⇒ Stop AAA⇒ TAA			zero				Female	Normal		Marcelli et al: Mol Endocrinol 4: 1105, 1990
0975	CAIS	Substitut.	3 DBD		593 2141	Tyr⇒ Stop TAC⇒ TAA	18	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0887	Prostate Cancer	Substitut.	3 DBD		594 2142	Leu⇒ Met CTG⇒ ATG						Q58L,Q260X,AQ86, E665D. Bical treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0053	PAIS	Substitut.	3 DBD	*	* 596 2148	Ala⇒ Thr GCC⇒ ACC			normal	normal		Found in 2 unrelated fam. Abolishes dimerization	Male	Ambiguous	pos	Gast et al; Mol & Cell Endocrinol 111: 93-98, 1995
0434	PAIS	Substitut.	3 DBD		* 596 2148	Ala⇒ Thr GCC⇒ ACC			normal	normal		Somatic mosaicism	Male	Ambiguous		Holterhus et al; Pediatric Res 46: 684 -690, 1999
0510	PAIS	Substitut.	3 DBD	*	* 596 2148	Ala⇒ Thr GCC⇒ ACC			normal			partial transactivation in COS cells	Male	Ambiguous		Giwercman et al; Hormone Research 53: 83-88, 2000
0710	PAIS	Substitut.	3 DBD		* 596 2148	Ala⇒ Thr GCC⇒ ACC			high				Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0959	E006AA Prost Can cell line	Substitut.	3 DBD		597 2148	Ser⇒ Gly AGC⇒ GGC						Dominant-negative loss of function mut AR gene amplificat	Male	Normal		D'Antonio et al. PLOS One 5:e11475, 2010
0054	PAIS	Substitut.	3 DBD	*	597 2151	Ser⇒ Gly AGC⇒ GGC			normal	normal		High dissoc. rate. Also has Arg617Pro (CGG toCCG) mut.	Female	Ambiguous		Zoppi et al; Mol Endocrinol 6:409, 1992
0390	PAIS	Substitut.	3 DBD		597 2152	Ser⇒ Thr AGC⇒ ACC						Servere hypospadia and cryptorchidism	Male	Ambiguous		Nordenskjold et al Urological Res. 27: 49 -55, 1999
0609	PAIS	Substitut.	3 DBD		597 2154	Ser⇒ Arg AGC⇒ AGA						Newborn treated with 1% DHT locally - normal penile size	Male	Ambiguous		Giwercmann et al; Horm Res 61: 58-62, 2004
0610	Normal	Substitut.	3 DBD		597 2154	Ser⇒ Arg AGC⇒ AGA						Adult had bilateral cryptorchadism in childhood	Male	Normal		Giwercmann et al; Horm Res 61: 58-62, 2004
0720	CAIS	Deletion	3 DBD		599 2158	Asn⇒ Met AΔAT⇒ ATG						single nt deletion causing frameshift & stop at codon 624	Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0055	CAIS	Substitut.	3 DBD		601 2164	Cys⇒ Phe TGC⇒ TTC							Female	Normal	pos	Baldazzi et al; Hum Mol Genet 3:1169-70 1994
0946	PAIS	Substitut.	3 DBD	*	601 2164	Cys⇒ Ser TGC⇒ TCC						40% loss of ligand-binding & 70% loss of transacrivation	Female	Ambiguous		Singh et al. J Steroid Biochem Mol Biol 2010
0362	PAIS	Substitut.	3 DBD		602 2166	Thr⇒ Pro ACT⇒ CCT	24					testis located in ingunal canal. Same family as 0831	Male	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0831	PAIS	Substitut.	3 DBD		602 2166	Thr⇒ Pro ACT⇒ CCT						Same family as 0362	Male	Ambiguous	pos	Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
0942	PAIS	Substitut.	3 DBD		603 2170	Ile⇒ Asn ATT⇒ AAT			zero			Somatic mosaicism - no transcriptional activity				Elfferich et al. Sexual Development 3:237 -244, 2009

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0056 PAIS	Substitut.	3 DBD			604 2172	Asp⇒Tyr GAT⇒TAT							Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
0598 PAIS	Substitut.	3 DBD			604 2172	Asp⇒Tyr GAT⇒TAT							Female	Ambiguous	pos	Scheiber et al; J Ped Endocrinol & Metab. 16: 367-373, 2003
0847 CAIS	Substitut.	3 DBD			607 2181	Arg⇒Stop CGA⇒TGA						Diag at 1Mo Bilateral gonadectomy at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
0848 CAIS	Substitut.	3 DBD		*	607 2181	Arg⇒Stop CGA⇒TGA						Diag at 1 Mo-Woolfian derivatives on rt side only	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
0057 CAIS	Substitut.	3 DBD		*	607 2181	Arg⇒Stop CGA⇒TGA			zero				Female	Normal		Brown et al; Eur J Pediatr (Suppl 2) 152; S62, 1993
0511 CAIS	Substitut.	3 DBD		*	607 2181	Arg⇒Stop CGA⇒TGA			zero				Female	Normal		Giwercman et al; Hormone Research 53: 83-88, 2000
0702 CAIS	Substitut.	3 DBD		*	607 2181	Arg⇒Stop CGA⇒TGA						1 affected sibling	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0770 MAIS	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0677 PAIS	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA								Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0393 PAIS	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA						Germ cell tumour - in undescended testis	Female	Normal		Chen et al; Human Reproduction 14: 664 -670, 1999
0347 PAIS	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA						Patient successfully treated with testostertone	Male	Ambiguous		Weidemann et al; J Clin Endocrinol & Metab 83: 1173-1176, 1998
0060 PAIS	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA							Female	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0059 PAIS	Substitut.	3 DBD	*	*	607 2182	Arg⇒Gln CGA⇒CAA			normal	normal			Male	Ambiguous	pos	Weidemann et al; Clin Endocrinology 45: 733 -739, 1996
0058 PAIS and breast cancer	Substitut.	3 DBD		*	607 2182	Arg⇒Gln CGA⇒CAA							Male	Ambiguous	pos	Wooster et al; Nat Genet 2:132, 1992
0412 CAIS	Substitut.	3 DBD			608 2184	⇒ ⇒						Mullerian ducts pres. 5nt. del frameshift & stop in codon 619	Female	Normal		Chen et al; Fertility & Sterility 72: 170-173, 1999
0061 PAIS	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG			normal	normal			Male	Ambiguous		Saunders et al; Clin Endocrinol 37:214, 1992
0062 PAIS and breast cancer	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG			normal	normal			Male	Ambiguous		Lobaccaro et al; Hum Mol Genet, 2:1799, 1993
0322 PAIS	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG			normal	normal		Defective nuclear localization	Male	Ambiguous		Tincello et al; Clinical Endocrinology 46: 497 -506, 1997
0352 PAIS	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG							Male	Ambiguous	pos	Hiort et al; J Pediatrics 132: 939- 943, 1998
0481 PAIS	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG			normal	high						Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0956 PAIS	Substitut.	3 DBD			608 2185	Arg⇒Lys AGG⇒AAG							Male	Ambiguous	neg	Wu et al. Fertility & Sterility 93:2076, e1-4, 2010
0063 PAIS	Substitut.	3 DBD	*		610 2190	Asn⇒Thr AAT⇒ACT			normal	low	*		Male	Ambiguous		Weidemann et al; Clin Endocrinology 45: 733 -
0721 CAIS	Insertion	3 DBD			610 2190	Asn⇒Lys AAT⇒AAAT							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0496 CAIS	Substitut.	3 DBD			611 2193	Cys⇒Tyr TGT⇒TAT							Female	Normal		Mockel et al; Geburtshi. und Frauen. 60: 232 -234, 2000
0722 CAIS	Substitut.	3 DBD			614 2203	Cys⇒Tyr TGT⇒TAT							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005

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0064	CAIS	Deletion	3 DBD		615 2204-6	Arg⇒0 TGATCG⇒TGT	27	23	normal	normal		3 nt. del -Arg615 del, 1nt. from 614, 2nt. 615. 614 still Cys	Female	Normal		Beitel et al; Hum Mol Genet, 3:21, 1994
0512	CAIS	Substitut.	3 DBD	*		615 2205	Arg⇒Gly CGT⇒GGT					no transactivation in COS cells	Female	Normal		Giwercman et al; Hormone Research 53: 83-88, 2000
0065	CAIS	Substitut.	3 DBD	*	*	615 2206	Arg⇒His CGT⇒CAT	25	23	low	high		Female	Normal	pos	Beitel et al; Hum Mol Genet, 3:21, 1994
0066	CAIS	Substitut.	3 DBD	*	*	615 2206	Arg⇒His CGT⇒CAT			normal	normal		Female	Normal	pos	Mowszowicz et al; Mol Endocrinol 7:861-869, 1993
0067	CAIS	Substitut.	3 DBD	*	*	615 2206	Arg⇒His CGT⇒CAT						Female	Normal		Brown et al; Eur J Pediatr 152 (Suppl 2): S62, 1993
0068	CAIS	Substitut.	3 DBD	*	*	615 2206	Arg⇒His CGT⇒CAT					2 affected siblings variable phenotype	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
0348	CAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT						Female	Normal		Cabral et al; Brazilian J Mol & Biol Res. 31: 775-778, 1998
0353	CAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT						Female	Normal		Hiort et al; J Pediatrics 132: 939- 943, 1998
0354	CAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT						Female	Normal		Hiort et al; J Pediatrics 132: 939- 943, 1998
0069	PAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT						Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0654	CAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT					Woolfian remnants present - sister of 655	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0655	CAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT					Sister of 654	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0069	MAIS	Substitut.	3 DBD		*	615 2206	Arg⇒His CGT⇒CAT					Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0070	PAIS	Substitut.	3 DBD			615 2206	Arg⇒Pro CGT⇒CCT						Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0445	CAIS	Substitut.	3 DBD			615 2206	Arg⇒Pro CGT⇒CCT			normal	high		Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0071	PAIS	Substitut.	3 DBD	*		616 2209	Leu⇒Arg CTT⇒CGT			normal	normal		Female	Ambiguous	pos	De Bellis et al; J Clin Endocrinol Metab, 78:513, 1994
0072	CAIS	Substitut.	3 DBD			616 2209	Leu⇒Pro CTT⇒CCT						Female	Normal		Mebarki et al; 75th US Endo Soc Meeting, Abstr 602,1993
0073	CAIS	Substitut.	3 DBD	*		616 2209	Leu⇒Pro CTT⇒CCT			normal	normal		Female	Normal		Lobaccaro et al; Mol Cell Endorinol, 5: 137 -147, 1996
0074	PAIS	Substitut.	3 DBD	*		617 2212	Arg⇒Pro CGG⇒CCG			normal	normal		Female	Ambiguous	pos	Marcelli et al; J Clin Invest. 87: 1123, 1991
0075	PAIS	Substitut.	3 DBD	*		617 2212	Arg⇒Pro CGG⇒CCG			normal	normal high	Mutation also at 597	Female	Normal		Zoppi et al; Mol Endocrinol 6:409, 1992
0431	Prostate cancer	Substitut.	3 DBD	*		619 2218	Cys⇒Tyr TGT⇒TAT			low	high	Inactive transcription Does not bind DNA somatic mutation	Male	Normal		Nazereth et al; Mol Endocrinol 13: 2065 -2075, 1999
0976	CAIS	Substitut.	3 DBD			619 2217	Cys⇒Arg TGT⇒CGT	20	18			Mother heterozygous carrier, 1 affected sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0491	Prostate cancer	Substitut.	3 DBD			619 2218	Cys⇒Tyr TGT⇒TAT					Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0076	CAIS	Deletion	3-8			⇒ ⇒							Female	Normal		Brown et al, Eur J Pediatr (Suppl 2) 152: S62, 1993
0077	MAIS	Deletion	4 LBD			⇒ ⇒						Azoospermia	Male	Normal	neg	Aiken et al; Am J Obs & Gyn . 165:1891 -1894, 1991

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0078	CAIS	Deletion	4 LBD	*	⇒ ⇒						13 nt deletion causing frameshift and stop at codon 783	Female	Normal	pos	Lobaccaro et al; Mol & Cellular Endocrinology 111: 21-8,1995
0711	PAIS	Substitut.	4		629 2247	Arg⇒Trp CGG⇒TGG			normal		40% higher transactivation than WT impaired N/C	Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0306	Prostate cancer	Substitut.	4		629 2248	Arg⇒Gln CGG⇒CAG					1 of 6 of hormone-independent D2 patients- somatic mut	Male	Normal		Wang et al; Japanese J of Urology 88: 550-556 1997
0079	Prostate cancer	Substitut.	4		630 2251	Lys⇒Thr AAG⇒ACG					Also Lys717Glu mut, (AAGtoGAG)+silent mut in 701. Som mut	Male	Normal		Tilley et al; Clinical Cancer Res. 2:277-285, 1996
0400	CAIS	Substitut.	4 LBD		640 2280	Gln⇒Stop CAG⇒TAG			zero			Female	Normal		Yaegashi et al; Tohoku J of Exp Med 187: 263 -272, 1999
0429	CAIS	Substitut.	4 LBD		640 2280	Gln⇒Stop CAG⇒TAG			zero		also Trp751Stop mut, (TGGtoTGA) 47XXY,	Female	Normal		Uehara et al; Am J Med Genet. 86: 107-111, 1999
0613	Prostate cancer	Substitut.	4 LBD	*	640 2280	Gln⇒Stop CAG⇒TAG					also T877A. transactiv androgen-independent~275x	Male	Normal		Ceraline et al. Int. J. Cancer 108: 152-157, 2004
0080	PAIS	Substitut.	4 LBD		645 2296	Ala⇒Asp GCT⇒GAT						Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0334	Normal	Substitut.	4 LBD		645 2296	Ala⇒Asp GCT⇒GAT						Male	Normal		Nordenskjold et al; Human Mutation. 11: 339, 1998
0617	CAIS	Substitut.	4 LBD		645 2296	Ala⇒Asp GCT⇒GAT	27		low		Also Arg752Gln	Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
0772	MAIS	Substitut.	4 LBD		645 2296	Ala⇒Asp GCT⇒GAT					male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0812	PAIS	Substitut.	4 LBD	*	645 2296	Ala⇒Asp GCT⇒GAT	28				Long poly Q and short poly G contribute to PAIS	Male	Ambiguous		Werner et al. J Clin Endocrinol Metab 91:3515-3520, 2006
0813	PAIS	Substitut.	4 LBD	*	645 2296	Ala⇒Asp GCT⇒GAT	30				Long poly Q and short poly G contribute to PAIS	Male	Ambiguous		Werner et al. J Clin Endocrinol Metab 91:3515-3520, 2006
0081	Prostate cancer	Substitut.	4 LBD		647 2302	Ser⇒Asn AGC⇒AAC					+ Gly724Asp,Leu880 Gln & Ala896Thr. mut	Male	Normal		Taplin et al; New England J Med 332:1393-1398, 1995
1020	Premature ovarian failure	Substitut.	4 LBD		649 2307	Thr⇒Ala ACC⇒GCC					Patient had menopause at 28	Female	Normal		Panda et al. Gyncol Endocrinol 2010
0576	Prostate cancer	Substitut.	4 LBD		649 2309	Thr⇒Thr ACC⇒ACT					Somatic mut. -CaP Poorly diff, Stage B2 +Estramustine treat	Male	Normal		Segawa et al; Int J of Urology 9: 545-553, 2002
0784	MAIS	Substitut.	4 LBD		650 2310	Ser⇒Gly AGC⇒GGC					Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0827	MAIS	Substitut.	4 LBD	*	650 2310	Ser⇒Gly AGC⇒GGC	23	18			Hypoandrogenism, scrotal hypoplsia hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68:58-588, 2008
0555	PAIS	Substitut.	4 LBD		653 2319	Glu⇒Lys GAG⇒AAG	20				Also in family with CAH with no androgen insensitivity	Male	Ambiguous		Lundberg et al; J Clin Endocrinol & Metab 87: 2023-2028, 2002
0517	CAIS	Substitut.	4 LBD		657 2331	Gln⇒Stop CAG⇒TAG						Female	Normal		Chavez et al; Clin Genet 59:: 185-188, 2001
1021	Premature ovarian failure	Substitut.	4 LBD		657 2331	Gln⇒Lys CAG⇒AAG					Patient had menopause at 17	Female	Normal		Panda et al. Gyncol Endocrinol 2010
0082	PAIS	Substitut.	4 LBD		664 2353	Ile⇒Asn ATT⇒AAT	22	22	low	norm					Pinsky et al; Clin Inv Med 15: 456, 1992
0937	Prostate cancer	Substitut.	4 LBD		665 2357	Glu⇒Asp GAA⇒					Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0083	Prostate cancer	Substitut.	4 LBD		670 2371	Gln⇒Arg CAG⇒CGG					Also Ser791Pro (TCT to CCT) mut. Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
0943	CAIS	Substitut.	4 LBD	*	671 2373	Pro⇒Ser CCC⇒TCC					Reduced transactivat no change in DNA or FxxLF binding	Female	Normal		Elfferich et al. Sexual Development 3:237 -244, 2009

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0084	PAIS	Substitut.		4	671	Pro⇒His							Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
		LBD		2374	CCC⇒CAC											
0085	Prostate cancer	Substitut.		4	672	Ile⇒Thr						Somatic mutation	Male	Normal		Tilley et al; Clinical Cancer Res. 2: 277-285, 1996
		LBD		2377	ATC⇒ACC											
0086	CAIS	Substitut.		4	677	Leu⇒Pro			zero				Female	Normal	pos	Belsham et al; Human Mutation 5: 28-33, 1995
		LBD		2392	CTG⇒CCG											
0723	CAIS	Substitut.		4	680	Ile⇒Thr						Affected twin sister	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
		LBD		2401	ATT⇒ACT											
1005	PAIS	Substitut.		4	680	Ile⇒Asn	21	4	normal	high		sister , cousin and 2 great aunts affected	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		LBD		2401	ATT⇒AAT											
0087	CAIS	Substitut.		4	681	Glu⇒Lys							Female	Normal		Hiort et al; J Clin Endocrinol Metab 77: 262-266, 1993
		LBD		2403	GAG⇒AAG											
0394	CAIS	Substitut.		4	681	Glu⇒Lys						Germ cell tumour in undescended testis	Female	Normal		Chen et al; Human Reproduction 14: 664 -670, 1999
		LBD		2403	GAG⇒AAG											
0618	CAIS	Substitut.		4	681	Glu⇒Stop	21		v low				Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
		LBD		2403	GAG⇒TAG											
0619	CAIS	Substitut.		4	681	Glu⇒Stop	21		low				Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
		LBD		2403	GAG⇒TAG											
947	CAIS	Substitut.		4	681	Glu⇒Asp						Mother a carrier- 3 affected siblings	Female	Normal	pos	Wu et al. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 26:606-609 2009
		LBD			GAG⇒GAT											
0534	PAIS	Substitut.		4	682	Pro⇒Thr			low				Female	Ambiguous		Chavez et al; J Hum Genet. 46: 560-565, 2001
		LBD		2406	CCA⇒ACA											
0724	CAIS	Substitut.		4	682	Pro⇒Ala			low			aunt, 2 sisters and neice affected	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
		LBD		2406	CCA⇒GCA											
0089	Prostate cancer	Substitut.		4	683	Gly⇒Ala						Somatic mutation - transactivation normal	Male	Normal		Koivisto et al; Cancer Research 57: 314-319, 1997
		LBD		2410	GGT⇒GCT											
0090	CAIS	Substitut.		4	684	Val⇒Ile			zero				Female	Normal		Mebarki et al; 75th US Endo Soc Meeting, Abstr 602, 1993
		LBD		2412	GTA⇒ATA											
0945	CAIS	Insertion		4	685	Val⇒						2 nt insert causing frameshift & stop in Codon 787	Female	Normal	pos	Rong et al; Eur J Obst & Gynecol & Reprod Biol 148:53-55, 2010
		LBD			GTG⇒GAGT											
0091	PAIS	Substitut.		4	686	Cys⇒Arg							Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
		LBD		2418	TGT⇒CGT											
0092	PAIS	Substitut.		4	687	Ala⇒Val							Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
		LBD		2422	GCT⇒GTT											
0093	CAIS	Substitut.		4	688	Gly⇒Glu						de novo mutation	Female	Normal	neg	Hiort et al; J Pediatrics 132: 939- 943, 1998
		LBD			GGA⇒											
0446	CAIS	Substitut.		4	688	Gly⇒Stop			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		LBD		2424	GGA⇒TGA											
0575	CAIS	Substitut.	*	4	689	His⇒Pro			v low	low	high	* v. low transactivation activity	Female	Normal		Rosa et al. J Clin Endocrinol & Metab 87; 4378-4382, 2002.
		LBD		2428	CAC⇒CCC											
0094	PAIS	Deletion		4	690	Asp⇒0										Schwartz et al; Horm Res 41:117 Abstr 244, 1994
		LBD		2430-2	GAC⇒0											
0656	CAIS	Substitut.	*	4	690	Asp⇒Val				high		Epidiymis & Vas deferens present - v low transactivation	Female	Normal		Tadokoro et al; Clinical Endocrinology 71:253 -260, 2009
		LBD		2431	GAC⇒GTC											
0095	CAIS	Deletion		4	692	Asn⇒0			normal	high	*	Three nucleotide deletion	Female	Normal		Batch et al; Hum Mol Genet 1:497, 1992
		LBD		2436-8	AAC⇒0											
0704	PAIS	Deletion		4	692	Asn⇒0			normal	high		3 nt. deletion. 3 affected siblings . variable phenotype	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
		LBD		2436-8	AAC⇒0											
0849	CAIS	Substitut.		4	693	Gln⇒Stop						Diag at 17y- Bilateral gonadectomy 17y	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD		2439	CAG⇒TAG											

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0096	CAIS	Substitut.	*	695 2445	Asp⇒His GAC⇒CAC			low				Female	Normal	neg	Ris-Stalpers et al; Mol Endocrinol 5:1562, 1991
0097	CAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC			normal	normal	high	mutation found in two unrelated families	Female	Normal	pos	Ris-Stalpers et al; Mol Endocrinol 5:1562, 1991
0098	PAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC						de novo mutation	Female	Ambiguous		Hiort et al; J Pediatrics 132: 939- 943, 1998
0657	CAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0773	MAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0977	CAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC						Diag at 2 wk bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
0978	CAIS	Substitut.	*	695 2445	Asp⇒Asn GAC⇒AAC	22	17				Mother & 1 cousin hetrozyg, 1 cousin & 1 aunt affect, sister wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0977	CAIS	Substitut.		695 2445	Asp⇒Tyr GAC⇒TAC	19	18				Mother heterozygous carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0335	CAIS	Substitut.		695 2446	Asp⇒Val GAC⇒GTC	21					mtuation found in two siblings	Female	Normal	pos	Dork et al; Human Mutation 11: 337-339, 1998
0447	CAIS	Substitut.		700 2460	Leu⇒Met TTG⇒ATG			zero			Epididymous and Vas deferns present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0725	CAIS	Deletion		700	Leu⇒0 TTG⇒						13 nt del framshift & stop at codon 783 sis & moth carr, neice aff	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
0448	CAIS	Substitut.		701 2463	Leu⇒Phe CTC⇒TTC							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0518	PAIS	Substitut.		701 2463	Leu⇒Ile CTC⇒ATC										Chavez et al; Clin Genet 59:: 185-188, 2001
0099	Prostate cancer	Substitut.		701 2464	Leu⇒His CTC⇒CAC						Somatic mutation	Male	Normal		Suzuki et al; J Steroid Biochem Molec Biol 46:759, 1993
0326	Prostate cancer	Substitut.		701 2464	Leu⇒His CTC⇒CAC						Somatic mutation	Male	Normal		Watanabe et al; Jpn J Clin Oncol 27: 389 -393, 1997
0408	MDA PCa-Za	Substitut.		701 2464	Leu⇒His CTC⇒CAC			normal	low		Som. mut. Prostate cancer cell line. Also has Thr877Ala	Male	Normal		Zao et al; J of Urology 162: 2192-2199, 1999
0100	CAIS	Substitut.		702 2466	Ser⇒Ala TCT⇒GCT			zero				Female	Normal		Pinsky et al; Clin Inv Med 15:456, 1992
0101	PAIS	Substitut.	*	703 2469	Ser⇒Gly AGC⇒GGC			low	high			Male	Ambiguous		Radnayr et al; J of Urology 158: 1553 -1556, 1997
0449	CAIS	Substitut.	*	703 2469	Ser⇒Gly AGC⇒GGC							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0712	PAIS	Substitut.		703 2469	Ser⇒Gly AGC⇒GGC				high			Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0713	PAIS	Substitut.		703 2469	Ser⇒Gly AGC⇒GGC							Female	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0979	CAIS	Substitut.		703 2469	Ser⇒Cys AGC⇒TGC	19	18	low	normal		Mother, sister, heterozygote carriers, I aunt wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0559	CAIS	Substitut.		705 2475	Asn⇒Tyr AAT⇒TAT						Sister a carrier	Female	Normal		Sills et al; Int J Mol Med 9: 45-48, 2002
0102	CAIS	Substitut.		705 2476	Asn⇒Ser AAT⇒AGT			zero				Female	Normal		Pinsky et al; Clin Inv Med 15:456, 1992
0103	CAIS	Substitut.		705 2476	Asn⇒Ser AAT⇒AGT			zero				Female	Normal		DeBellis et al; Mol Endocrinol 6:1909-20, 1992

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0104	CAIS	Substitut.		705	Asn⇒ Ser						Mutation found in two unrelated families	Female	Normal		Quigley et al; Endocrine Reviews 16: 271, 1995
		4 LBD		2476	AAT⇒ AGT										
0482	PAIS	Substitut.		705	Asn⇒ Ser			normal	high						Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		4 LBD		2476	AAT⇒ AGT										
0633	CAIS	Substitut.		705	Asn⇒ Ser			zero			Testes located in abdomen	Female	Normal	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		4 LBD		2476	AAT⇒ AGT										
0634	CAIS	Substitut.		705	Asn⇒ Ser	22		zero			Testes located in inguinal region	Female	Normal	pos	Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
		4 LBD		2476	AAT⇒ AGT										
0980	CAIS	Substitut.		705	Asn⇒ Ser	18	18	zero	zero			Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		4 LBD		2476	AAT⇒ AGT										
0981	CAIS	Substitut.		705	Asn⇒ Ser	22	18				1 affected sister, 1 heterozygote sister, 2 wt sisters	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		4 LBD		2476	AAT⇒ AGT										
0581	CAIS	Substitut.		705	Asn⇒ Ile						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
		4 LBD		2476	AAT⇒ ATT										
0582	CAIS	Substitut.		705	Asn⇒ Ile						Low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
		4 LBD		2476	AAT⇒ ATT										
0105	CAIS	Substitut.	*	707	Leu⇒ Arg							Female	Normal		Lumbroso et al; J Clin Endo & Metab 81: 1984 -1988, 1996
		4 LBD		2482	CTG⇒ CGG										
0106	PAIS	Substitut.		708	Gly⇒ Ala							Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
		4 LBD		2485	GGA⇒ GCA										
0314	PAIS	Substitut.		708	Gly⇒ Ala						Servere hypospadias	Male	Ambiguous		Albers et al; J of Pediatrics 131: 388 -392, 1997
		4 LBD		2485	GGA⇒ GCA										
0107	PAIS	Substitut.		708	Gly⇒ Val			zero			Mother had both mut. & wt. Ar genes.	Male	Ambiguous	pos	Auchus et al: 77th US Endo Soc Meeting, Abstr P1-508 1995
		4 LBD		2485	GGA⇒ GTA										
1023	CAIS	Substitut.		708	Gly⇒ Val			low			Almost complete loss of transaction	Female	Normal		Rajender et al. J Androl 2010
		4 LBD		2485	GGA⇒ GAA										
0819	PAIS	Substitut.	*	709	Glu⇒ Lys							Female	Ambiguous		Geoget et al. Mol Endocrinol 20:724-734, 2006
		4 LBD		2487	GAG⇒ AAG										
0577	Prostate cancer	Substitut.		709	Glu⇒ Glu						Somatic mut. CaP Moder diff, Stage D1 + biculitamide treat	Male	Normal		Segawa et al; Int J of Urology 9: 545-553, 2002
		4 LBD		2489	GAG⇒ GAA										
0658	CAIS	Substitut.		710	Arg⇒ Thr			zero			Sister to 0450 - Epididymis & Vas deferens present	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		4 LBD		2491	AGA⇒ ACA										
0450	CAIS	Substitut.		710	Arg⇒ Thr			zero			Sister to 0658- Epidiymis & Vas deferens present	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		4 LBD		2491	AGA⇒ ACA										
0525	PAIS	Substitut.	*	711	Gln⇒ Glu			v low			altered AR specificity 2x increased affinity for E2.	Female	Ambiguous	pos	Lumbroso et al. 83rd US Endo Soc Meeting, Abstr P2-29, 2001
		4 LBD		2493	CAG⇒ GAG										
0535	PAIS	Substitut.	*	711	Gln⇒ Glu			normal				Female	Ambiguous	pos	Chavez et al; J Hum Genet. 46: 560-565, 2001
		4 LBD		2493	CAG⇒ GAG										
0828	CAIS	Deletion		711	Gln⇒						I nt del-frameshift & stop in codon 788- 3 affected aunts	Female	Normal	pos	Galani et al; Fertility & Sterility 2008
		4 LBD		2494	CΔAG⇒ CGC										
0578	Prostate cancer	Substitut.		711	Gln⇒ Gln						Somatic mut. CaP Moder diff, Stage D2 + leuprolide treat	Male	Normal		Segawa et al; Int J of Urology 9: 545-553, 2002
		4 LBD		2495	CAG⇒ CAA										
0108	PAIS	Substitut.	*	712	Leu⇒ Phe			normal	high		Phenotypic diversity brother of 505& 506 Testost-induced	Male	Ambiguous	pos	Hiort et al; Am J Med Genet. 63: 218-222, 1996
		4 LBD		2496	CTT⇒ GTT										
0505	PAIS	Substitut.	*	712	Leu⇒ Phe			normal	high		Phenotypic diversity brother of 108& 506 Testost-induced	Male	Ambiguous	pos	Hiort et al; J Clin Endocrinol & Metab 85: 3245-3250, 2000
		4 LBD		2496	CTT⇒ GTT										
0506	PAIS	Substitut.	*	712	Leu⇒ Phe			normal	high		Phenotypic diversity brother of 505& 108 Testost-induced	Male	Ambiguous	pos	Hiort et al; J Clin Endocrinol & Metab 85: 3245-3250, 2000
		4 LBD		2496	CTT⇒ GTT										
0507	PAIS	Substitut.	*	712	Leu⇒ Phe			normal	high		Phenotypic diversity uncle of 108,505,506 Testost-induced	Male	Ambiguous	pos	Hiort et al; J Clin Endocrinol & Metab 85: 3245-3250, 2000
		4 LBD		2496	CTT⇒ GTT										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot		Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0109 Prostate cancer	Substitut.	4 LBD	*	*	715 2507	Val⇒Met GTG⇒ATG			normal			Somatic mutation. Receptor showed a gain in function	Male	Normal		Culig et al; Mol Endocrinol 7:1541-1550 1993
0110 Prostate cancer	Substitut.	4 LBD		*	715 2507	Val⇒Met GTG⇒ATG			normal			Somatic mutation. Receptor showed a gain in function	Male	Normal		Bubley et al 87th Am Assoc Cancer Res Meet Abstr. 1680, 1996
0678 CAIS	Substitut.	4 LBD		*	715 2507	Val⇒Stop GTG⇒						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0792 PAIS	Substitut.	4 LBD			718 2514	Trp⇒Arg TGG⇒CGG						Somatic mosaic 2/3 mutant to 1/3 wt				Mueller et al. Hum Genet 119:680, 2006
0111 CAIS	Substitut.	4 LBD			718 2516	Trp⇒Stop TGG⇒TGA			zero				Female	Normal	pos	Sai et al; Am J Hum Genet 46:1095, 1990
0112 Prostate cancer	Substitut.	4 LBD			720 2520	Lys⇒Glu AAG⇒GAG						Somatic mutation- Bone metasteses of Prostate cancer	Male	Normal		Kleinerman et al; J of Urology 155: 624A, 1996
0113 Prostate cancer	Substitut.	4 LBD			721 2523	Ala⇒Thr GCC⇒ACC						Somatic mutation in 20% of isolates in initial cloning	Male	Normal		Taplin et al; New England J Med 332: 1393-1398, 1995
0583 CAIS	Deletion	4 LBD			721 2523	Ala⇒0 GΔCC⇒GTT						2nt.frameshift & stop in codon 766 - no immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0584 CAIS	Deletion	4 LBD			721 2523	Ala⇒0 GΔCCT⇒TGC						3nt.frameshift - low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0114 CAIS	Substitut.	4 LBD			722 2526	Leu⇒Phe TTG⇒TTT							Female	Normal		Hiort et al: Am J Med Genet. 63: 218-222, 1996
0761 Prostate cancer	Substitut.	4 LBD			722 2526	Leu⇒Phe TTG⇒TTT						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0451 CAIS	Substitut.	4 LBD			723 2529	Pro⇒Ser CCT⇒TCT			normal high			-Epidiymis present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0791 PAIS ?	Substitut.	4 LBD			723 2528	Pro⇒Leu CCT⇒CTT										Mueller et al. Hum Genet 119:681, 2006
0452 CAIS	Substitut.	4 LBD			724 2532	Gly⇒Ser GGC⇒AGC			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0453 CAIS	Substitut.	4 LBD			724 2533	Gly⇒Asp GGC⇒GAT			zero			Epidimis & Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0620 CAIS	Substitut.	4 LBD			724 2533	Gly⇒Ala GGC⇒GCC	19		normal				Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
0798 CAIS	Substitut.	4 LBD	*		724 2533	Gly⇒Val GGC⇒GTC			zero			Severley impaired AR transactivation	Female	Normal		Jaaskelainen et al. Hum Mutat 27:291, 2006
0115 CAIS	Deletion	4-8 LBD				⇒ ⇒			zero				Female	Normal		Brown et al; Proc Natl Acad Sci 85:8151, 1988
0116 CAIS	Deletion	5 LBD				⇒ ⇒			zero			Affected aunt deleted for exons 6 and 7 only.	Female	Normal	pos	Maclean et al; J Clin Invest, 91:1123, 1993
0117 CAIS	Substitut.	5 LBD				Tyr⇒Arg ⇒			zero				Female	Normal		Marcelli et al; 74th US Endo Soc Meetings: Abstr. 224, 1992
0118 PAIS	Substitut.	5 LBD	*		725 2535	Phe⇒Leu TTC⇒CTC			normal normal			Mutation disrupts N/C terminal interaction	Male	Ambiguous	pos	Quigley et al; Mech of Aging & Develop 125: 683-695, 2004
0391 PAIS	Substitut.	5 LBD			725 2535	Phe⇒Leu TTC⇒CTC						Hypospadia and cryptorchidism	Male	Ambiguous	pos	Nordenskjold et al Urological Res, 27: 49 -55, 1999
0119 Prostate cancer	Substitut.	5 LBD	*		726 2539	Arg⇒Leu CGC⇒CTC			normal normal			Germ line mutation present in offspring	Male	Normal	pos	Elo et al ; J Clin Endorinol Metab, 80: 3494-3500, 1995
0508 Prostate cancer	Substitut.	5 LBD	*		726 2539	Arg⇒Leu CGC⇒CTC						Estimated that 2% of Finnish CAP patients have this mutation	Male	Normal	pos	Mononen et al; Cancer Res 60: 6479-6481, 2000
0571 Prostate Cancer	Substitut.	5 LBD			726 2539	Arg⇒Leu CGC⇒CTC	20					Somatic mut polyGln 24 to 20 Orchiectomy Horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change Amino acid Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0687 Normal	Substitut.	5 LBD		726 2539	Arg⇒Leu CGC⇒CTC							Patient suffereing frm alcoholism		Normal		Yan et al; Psychiatric Genet 14; 57-60, 2004
0120 MAIS	Substitut.	5 LBD		727 2543	Asn⇒Lys AAC⇒AAG							Oligospermia	Male	Normal		Yong et al; Lancet, 344: 826-827, 1994
0121 PAIS	Substitut.	5 LBD		728 2545	Leu⇒Ser TTA⇒TCA			low			*					McPhaul et al; J Clin Inv, 90:2097, 1992
0122 Prostate Cancer	Substitut.	5 LBD	*	730 2550	Val⇒Met GTG⇒ATG							Somatic mutation	Male	Normal		Newmark et al; Proc Natl AcadSci 89:6319, 1992
0123 Prostate Cancer	Substitut.	5 LBD	*	730 2550	Val⇒Met GTG⇒ATG							Somatic mutation	Male	Normal		Petersiel et al; Int J Cancer 63: 544-550, 1995
0762 Prostate Cancer	Substitut.	5 LBD	*	730 2550	Val⇒Met GTG⇒ATG							patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0125 CAIS	Substitut.	5 LBD	*	732 2556	Asp⇒Tyr GAC⇒TAC				high				Female	Normal		Brown et al; 74th US Endo Soc Meeting, Abstr 1506, 1992
0126 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Tyr GAC⇒TAC			zero					Female	Normal		Pinsky et al; Clin Inv Med 15:456, 1992
0127 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Tyr GAC⇒TAC								Female	Normal		Ghirri and Brown; Pediatr Res 33: Abstr 95, 1993
0801 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Tyr GAC⇒TAC							6 affected family members	Female	Normal	pos	Scott et al. Endocr Pract 12:664-669, 2006
0124 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Asn GAC⇒AAC				high				Female	Normal		Brown et al; 74th US Endo Soc Meeting, Abstr 1506, 1992
0659 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Asn GAC⇒AAC							Epididymis & Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0310 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Asn GAC⇒AAC	19							Female	Normal		Ko et al; J Reprod. Med 42: 424- 427, 1997
0955 CAIS	Substitut.	5 LBD		732 2556	Asp⇒Asn GAC⇒AAC								Female	Normal	neg	Wu et al. Fertility & Sterility 93:2076, e1-4, 2010
0628 Prostate cancer	Substitut.	5 LBD		732 2558	Asp⇒Asp GAC⇒GAT							Orch + Bicalutiamide +Iphosphamide treat Gleason 10. Somatic	Male	Normal		Haaplaa et al. Lab Invest. 81:1647-1651, 2001
0752 CAIS	Substitut.	5 LBD		733 2559	Gln⇒Stop CAG⇒TAG							No WD development	Female	Normal	neg	Barbaro et al. Clin Endocrinol 66:822-826, 2007
0128 PAIS	Substitut.	5 LBD		733 2561	Gln⇒His CAG⇒CAT							This patient was a mosaic for wt. & mut. alleles- de novo mut.	Female	Ambiguous	neg	Hiort et al; J Pediatrics 132: 939- 943, 1998
0726 CAIS	Substitut.	5 LBD		737 2571	Ile⇒Phe ATT⇒TTT								Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0129 PAIS	Substitut.	5 LBD	*	737 2572	Ile⇒Thr ATT⇒ACT			low				Mutation disrupts N/C terminal interaction	Male	Ambiguous	pos	Quigley et al; Mech Aging & Develop 125:683-695
0944 CAIS	Substitut.	5 LBD	*	738 2575	Gln⇒Arg CAG⇒CGG							V reduced transactiv reduced interaction with FxxLX	Female	Normal		Elfferich et al. Sexual Development 3:237 -244, 2009
0530 CAIS	Substitut.	5 LBD	*	739 2577	Tyr⇒Asp TAC⇒GAC			zero				no transactivation in COS-1 cells	Female	Normal		Suzuki et al. Int. J Andrology 24: 183-188, 2001
0616 PAIS	Substitut.	5 LBD	*	740 2581	Ser⇒Cys TCC⇒TGC			v low				Acute stress masked PAIS.	Male	Ambiguous	pos	Pitteloud et al. J Clin Endorinol & Metab 89:1053-1058, 2004
0982 CAIS	Substitut.	5 LBD		741 2583	Trp⇒Arg TGG⇒CGG	23	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0130 CAIS	Substitut.	5 LBD	*	741 2583	Trp⇒Arg TGG⇒CGG			low					Female	Normal	neg	Marcelli et al; J Clin Invest 94: 1642-1650, 1994
0360 Prostate cancer	Substitut.	5 LBD		741 2584	Trp⇒Stop TGG⇒TAG							Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0595	CAIS	Substitut.	*	741	Trp⇒Leu							Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
		5 LBD		2584	TGG⇒TTG											
0858	CAIS	Substitut.		741	Trp⇒Leu							Diag at birth bilateral gonadectomy at 14yr	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		5 LBD		2584	TGG⇒TTG											
0859	CAIS	Substitut.		741	Trp⇒Leu							Diag at birth bilateral gonadectomy at 12.5yr	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		5 LBD		2584	TGG⇒TTG											
0629	Prostate cancer	Substitut.		741	Trp⇒Cys							Orch + bicalutamide Gleason 10- somatic mutation	Male	Normal		Haaplaa et al. Lab Invest. 81:1647-1651, 2001
		5 LBD		2585	TGG⇒TGT											
0635	PAIS	Substitut.		741	Trp⇒Cys	27		low	high			Testes located in scrotum - Also has Leu547Phe mutation	Male	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		5 LBD		2585	TGG⇒TGT											
0552	Prostate cancer	Substitut.		741	Trp⇒Cys							Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al; J Clinical Oncology 21: 2673 -2678, 2003
		5 LBD		2585	TGG⇒TGT											
0131	PAIS	Substitut.		742	Met⇒Val					high						Ris-Stalpers et al; Pediatric Res. 36: 227 -234,1994
		5 LBD		2586	ATG⇒GTG											
0341	PAIS	Substitut.		742	Met⇒Val	21		zero				Testis located in inginual region	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		5 LBD		2586	ATG⇒GTG											
0744	PAIS	Substitut.		742	Met⇒Val							affected neice of 0131	Female	Ambiguous		Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
		5 LBD		2586	ATG⇒GTG											
0132	PAIS	Substitut.	*	742	Met⇒Ile			normal	high		*		Female	Ambiguous		Batch et al; Hum Mol Genet 1:497, 1992
		5 LBD		2588	ATG⇒ATA											
0948	PAIS	Substitut.		742	Met⇒Ile								Female	Ambiguous		Nagaraja et al. J Pediatr Endocrinol Metab 22:1169-1173, 2009
		5 LBD		2588	ATG⇒ATA											
1006	PAIS	Substitut.		742	Met⇒Ile	18	18	low	normal			Mother heterozygote carrier	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		5 LBD		2588	ATG⇒ATA											
0519	CAIS	Substitut.		743	Gly⇒Arg								Female	Normal		Chavez et al; Clin Genet 59:: 185-188, 2001
		5 LBD		2589	GGG⇒CGG											
0983	CAIS	Substitut.		743	Gly⇒Val	24	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		5 LBD		2590	GGG⇒GTG											
0133	PAIS	Substitut.	*	743	Gly⇒Val			low		high		Transcription only at high conc of androgen	Female	Ambiguous		Georget et al; J Clin Endocrinol & Metab 83: 3597-3603, 1998
		5 LBD		2590	GGG⇒GTG											
0134	PAIS	Substitut.		743	Gly⇒Val			normal	normal		*					Nakao et al; J Clin Endocrinol Metab 77:103-107, 1993
		5 LBD		2590	GGG⇒GTG											
0414	CAIS	Substitut.		743	Gly⇒Val			zero				de novo mutation	Female	Normal		Lobaccaro et al; J Steroid Biochem & Mol Biol. 44: 211-216, 1993
		5 LBD		2590	GGG⇒GTG											
0536	CAIS	Substitut.		743	Gly⇒Glu			normal					Female	Normal		Chavez et al; J Hum Genet. 46: 560-56, 2001
		5 LBD		2590	GGG⇒GAG											
0564	CAIS	Substitut.	*	743	Gly⇒Glu			v low	high	high	*		Female	Normal		Pujol et al; J Clin Endorinol & Metab. 87: 5793-5800. 2002
		5 LBD		2590	GGG⇒GAG											
0572	Prostate cancer	Substitut.		743	Gly⇒Gly							Somatic mutation + estrogen treatment Horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
		5 LBD		2591	GGG⇒GGC											
0361	Prostate cancer	Deletion		743	Gly⇒Gly							Frameshift-somatic mut.- separate tumor in same indv. as 0362	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2591	GGΔG⇒GGC											
0135	CAIS	Substitut.		744	Leu⇒Phe							1 affected sibling			pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
		5 LBD		2592	CTC⇒TTC											
0984	CAIS	Substitut.		744	Leu⇒Phe	21	17					1 heterozygote sister & 1 wt sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		5 LBD		2592	CTC⇒TTC											
0362	Prostate cancer	Substitut.		744	Leu⇒Phe							Somatic mutation - separate tumor in same indv. as 0361	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2592	CTC⇒TTC											
1007	PAIS	Substitut.		745	Met⇒Leu	23	17					mother & grandmother heterozyote carriers	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		5 LBD		2595	ATG⇒CTG											

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0136	PAIS	Substitut. 5 LBD		745 2596	Met⇒Thr ATG⇒ACG			zero							Ris-Stalpers et al; Pediatric Res 36: 227 -234, 1994
0727	CAIS	Substitut. 5 LBD		745 2596	Met⇒Thr ATG⇒ACG						Mother and sisters heterozygotous	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
0755	CAIS	Substitut. 5 LBD	*	745 2597	Met⇒Ile ATG⇒ATC						Enanced affinity for & increased transact for estradiol	Female	Normal		Bonagura et al. Mol Cell Endocrinol 263:79 -89, 2007
0138	PAIS	Substitut. 5 LBD		746 2598	Val⇒Met GTG⇒ATG							Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0137	PAIS	Substitut. 5 LBD		746 2598	Val⇒Met GTG⇒ATG										Brown et al; 74th US Endo Soc Meeting, Abstr 1506, 1992
0949	PAIS	Substitut. 5 LBD		746 2598	Val⇒Met GTG⇒ATG										Nagaraja et al. J Pediatr Endocrinol Metab 22:1169-1173, 2009
0785	MAIS	Substitut. 5 LBD		747 2601	Phe⇒Ile TTT⇒ATT						Male Infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0985	CAIS	Substitut. 5 LBD		747 2602	Phe⇒Cys TTT⇒TGT	23	17				1 wt sister	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0763	Prostate cancer	Deletion 5 LBD		747 2603	Phe⇒Leu TTΔT⇒TTG						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0492	Prostate cancer	Substitut. 5 LBD		748 2604	Ala⇒Thr GCC⇒ACC						Also Ser865Pro; Gln867Stop and Gln919Arg ;Som mut	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0565	Prostate cancer	Substitut. 5 LBD	*	748 2604	Ala⇒Thr GCC⇒ACC			normal	high		Somatic mutation. weaker interaction with HSP	Male	Normal		James et al; Mol Endocrinol. 16: 2692 -2705, 2002
0139	PAIS	Substitut. 5 LBD	*	748 2605	Ala⇒Asp GCC⇒GAC			low	high		Abnormal dissociation				Marcelli et al; J Clin Invest 94: 1642-1650, 1994
0363	Prostate cancer	Substitut. 5 LBD		748 2605	Ala⇒Val GCC⇒GTC						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0140	CAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG							Female	Normal	pos	DeBellis et al; Mol Endocrinol 6:1909-20, 1992
0141	CAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG							Female	Normal	pos	Jakubiczka et al; Hum Genet 90:311-2, 1992
0483	PAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG			normal	high						Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0614	CAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG						Fallopian tube present	Female	Normal		Van et al. Eur J. Pediatr. 162: 781-784, 2003
0679	CAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0680	CAIS	Substitut. 5 LBD		749 2607	Met⇒Val ATG⇒GTG						Diff between ext genital & chrom - in amniocentesis	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0364	Prostate cancer	Substitut. 5 LBD		749 2609	Met⇒Ile ATG⇒ATA						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0630	Prostate cancer	Substitut. 5 LBD		749 2609	Met⇒Ile ATG⇒ATA						Orch + bicalutamide Gleason 10 +silent mut Gln867Gln	Male	Normal		Haaplaa et al. Lab Invest. 81:1647-1651, 2001
0365	Prostate cancer	Substitut. 5 LBD		750 2610	Gly⇒Ser GGC⇒AGC						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0142	CAIS	Substitut. 5 LBD	*	750 2611	Gly⇒Asp GGC⇒GAC			v low			Mutation found in two unrelated patients	Female	Normal		Bevan et al; J Steroid Biochem Molec. Biol 61: 19-26, 1997
0143	CAIS	Substitut. 5 LBD		750 2611	Gly⇒Asp GGC⇒GAC							Female	Normal		Brown et al: 74th US Endo Soc Meeting Abstr 1506, 1992
0703	CAIS	Substitut. 5 LBD		751 2613	Trp⇒Arg TGG⇒AGG			normal	high		Sibling of 0144	Female	Normal		Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Androgen Binding: Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0144	CAIS	Substitut.		751	Trp⇒Arg			low	high		Sibling of 0703	Female	Normal	pos	Brinkmann et al; J Steroid Biochem Mol Biol 53: 443, 1995
		5 LBD		2613	TGG⇒AGG										
0366	Prostate cancer	Substitut.		751	Trp⇒Stop						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2614	TGG⇒TAG										
0367	Prostate cancer	Substitut.		751	Trp⇒Stop						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2614	TGG⇒TAG										
0368	Prostate cancer	Substitut.		751	Trp⇒Stop						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2615	TGG⇒TGA										
0401	CAIS	Substitut.		751	Trp⇒Stop			zero				Female	Normal		Yaegashi et al; Tohoku J of Exp Med 187: 263 -272, 1999
		5 LBD		2615	TGG⇒TGA										
0664	PAIS	Substitut.		751	Trp⇒Stop						Somatic mosaicism - male changed to female at 18mths	Female	Ambiguous		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
		5 LBD		2615	TGG⇒TGA										
0145	CAIS	Substitut.	*	752	Arg⇒Stop			zero				Female	Normal		Pinsky et al; Clin Inv Med 15:456, 1992
		5 LBD		2616	CGA⇒TGA										
0146	CAIS	Substitut.	*	752	Arg⇒Stop							Female	Normal		Brinkmann et al; J Steroid Biochem Mol Biol 53: 443, 1995
		5 LBD		2616	CGA⇒TGA										
0342	CAIS	Substitut.	*	752	Arg⇒Stop	16		zero			Testes located in abdomen	Female	Normal	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		5 LBD		2616	CGA⇒TGA										
0402	CAIS	Substitut.	*	752	Arg⇒Stop			zero				Female	Normal		Yaegashi et al; Tohoku J of Exp Med 187: 263 -272, 1999
		5 LBD		2616	CGA⇒TGA										
0728	CAIS	Substitut.	*	752	Arg⇒Stop							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
		5 LBD		2616	CGA⇒TGA										
0754	CAIS	Substitut.	*	752	Arg⇒Gln						Inherted from maternal grandmother present in two sisters	Female	Normal	pos	Olafsson et al. Laeknabladid 86:263 -166, 2000
		5 LBD		2617	CGA⇒CAA										
0147	CAIS	Substitut.	*	752	Arg⇒Gln			zero			Mutation found in two unrel. families. Equivalent to tfm rat	Female	Normal		Brown et al; 74th US Endo Soc Meeting, Abstr 1506, 1992
		5 LBD		2617	CGA⇒CAA										
0333	CAIS	Substitut.	*	752	Arg⇒Gln							Female	Normal	pos	Komori et al; Arch Gynecol & Obstetrics 261: 95-100, 1998
		5 LBD		2617	CGA⇒CAA										
0349	CAIS	Substitut.	*	752	Arg⇒Gln							Female	Normal		Cabral et al; Brazilian J Med & Biol Res. 31: 775-758, 1998
		5 LBD		2617	CGA⇒CAA										
0148	CAIS	Substitut.	*	752	Arg⇒Gln			zero			Equivalent to tfm rat	Female	Normal		Evans; J Endocrinol 135 Suppl, Abstr P26, 1992
		5 LBD		2617	CGA⇒CAA										
0497	CAIS	Substitut.	*	752	Arg⇒Gln						Bilateral testicular tumors	Female	Normal		Sakai et al; IntJ of Urology 7: 390-392, 2000
		5 LBD		2617	CGA⇒CAA										
0860	CAIS	Substitut.	*	752	Arg⇒Gln						Prenatal Diagnosis	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
		5 LBD		2617	CGA⇒CAA										
0879	CAIS	Substitut.	*	754	Phe⇒Val				high		Greatly reduced transactivation 5%	Female	Normal		Tadokoro et al; Clinical Endocrinology 71:253 -260, 2009
		5 LBD		2622	TTC⇒GTC										
0150	CAIS	Substitut.		754	Phe⇒Val							Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
		5 LBD		2622	TTC⇒GTC										
0149	CAIS	Substitut.		754	Phe⇒Val			zero				Female	Normal		Lobaccaro et al; Hum Mol Genet 2:1041 -1043, 1993
		5 LBD		2622	TTC⇒GTC										
0880	PAIS	Substitut.	*	754	Phe⇒Ser				normal		Microphallus no hypospadias	Male	Ambiguous		Tadokoro et al; Clinical Endocrinology 71:253 -260, 2009
		5 LBD		2623	TTC⇒TCC										
0369	Prostate cancer	Substitut.		754	Phe⇒Leu						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		5 LBD		2622	TTC⇒CTC										
0715	PAIS	Substitut.	*	754	Phe⇒Leu				high		Microphallus, hypospadias, cryptorchisms	Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
		5 LBD		2623	TTC⇒TTC										
0151	PAIS	Substitut.		754	Phe⇒Leu							Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
		5 LBD		2624	TTC⇒TTA										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain		CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0152 PAIS	Substitut.	5 LBD	*		754 2624	Phe⇒Leu TTC⇒TTA			normal	high		*		Male	Ambiguous		Weidemann et al; Clin Endocrinology 45: 733 -739, 1996
0714 PAIS	Substitut.	5 LBD			754 2624	Phe⇒Leu TTC⇒TTA				high		*		Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0370 Prostate cancer	Substitut.	5 LBD			755 2625	Thr⇒Ala ACC⇒GCC							Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0602 Prostate cancer	Substitut.	5 LBD			756 2628	Asn⇒Asp AAT⇒GAT							Flutamide treated - somatic mutation	Male	Normal		Taplin et al; J Clinical Oncology 21: 2673 -2678, 2003
0153 PAIS	Substitut.	5 LBD			756 2629	Asn⇒Ser AAT⇒AGT								Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0532 MAIS	Substitut.	5 LBD	*		756 2629	Asn⇒Ser AAT⇒AGT				high			Severe oligospermia- transactivation 38% of wt.	Male	Normal		Giwercman et al. Clin Endocrinol 54: 827 -834, 2001
0573 Prostate cancer	Substitut.	5 LBD			757 2631	Val⇒Ile GTC⇒ATC							Somatic mutation + Orchiectomy Horm-refractory CaP	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
0300 Prostate cancer	Substitut.	5 LBD	*		757 2632	Val⇒Ala GTC⇒GCC							Binds R1881 norm.- transcriptionally inactive- Som mut	Male	Normal		James et al; 79th US Endo Soc Meeting, Abstr P2-484, 1997
0493 Prostate cancer	Substitut.	5 LBD			757 2632	Val⇒Ala GTC⇒GCC							Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0346 PAIS	Substitut.	5 LBD	*		758 2635	Asn⇒Thr AAC⇒ACC			normal		high	*	50% reduction in transactivation in COS-7				Yong et al; Mol & Cell Endocrinol. 137: 41-50, 1998
0371 Prostate cancer	Substitut.	5 LBD			759 2637	Ser⇒Pro TCC⇒CCC							Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0154 CAIS	Substitut.	5 LBD			759 2638	Ser⇒Phe TCC⇒TTC			zero					Female	Normal		DeBellis et al; Mol Endocrinol, 6:1909-20, 1992
0605 CAIS	Deletion/ insertion	5 LBD			760 2640	Arg⇒0 ⇒			zero				7bp del (2640-2646) 11bp ins (2652-2662) stop 9 codon downstr.	Female	Normal	pos	Vichlis et al. J Hum Genet 48:346-351, 2003
0841 PAIS	Substitut.	5 LBD			760	Arg⇒Ser AGG⇒											Jeske et al; J Pediatr Endocrinol Metab 20:893-908, 2007
0938 Prostate cancer	Substitut.	5 LBD			760 2642	Arg⇒Arg AGG⇒							Treated with anti-androgens. Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0729 PAIS	Substitut.	5 LBD			761 2644	Met⇒Thr ATG⇒ACG							mother heterozygous	Male	Ambiguous	pos	Ledig et al: Horm Res 63:263-269, 2005
0155 CAIS	Substitut.	5 LBD			762 2646	Leu⇒Phe CTC⇒TTC			zero					Female	Normal		Brown et al; 74th US Endo Soc Meeting, Abstr 1506, 1992
0156 CAIS	Substitut.	5 LBD	*		762 2646	Leu⇒Phe CTC⇒TTC			zero					Female	Normal		Bevan et al; J Steroid Biochem Molec. Biol 61: 19-26, 1997
0861 CAIS	Substitut.	5 LBD			762 2646	Leu⇒Phe CTC⇒TTC							Diag at 7yr bilateral gonadectomy at 8yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
0862 CAIS	Substitut.	5 LBD			762 2646	Leu⇒Phe CTC⇒TTC							Diagnosed at 1 wk	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
0157 CAIS	Substitut.	5 LBD			763 2649	Tyr⇒His TAC⇒CAC								Female	Normal		Quigley et al; Endocrin. Reviews, 16:271, 1995
0159 PAIS	Substitut.	5 LBD			763 2650	Tyr⇒Cys TAC⇒TGC			low					Male	Ambiguous		Morono et al; Human Mutation 6: 152-162, 1995
0405 PAIS	Substitut.	5 LBD			763 2650	Tyr⇒Cys TAC⇒TGC								Male	Ambiguous		Batch et al; Arch Disease Child 68: 453, 1993
0484 PAIS	Substitut.	5 LBD			763 2650	Tyr⇒Cys TAC⇒TGC			normal	high							Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0485 PAIS	Substitut.	5 LBD			763 2650	Tyr⇒Cys TAC⇒TGC			normal	high							Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot		Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0636	PAIS	Substitut.	5		763	Tyr⇒Cys	22		low				Gynocomastia -testis located in scrotum	Male	Ambiguous	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		LBD			2650	TAC⇒TGC											
0372	Prostate Cancer	Substitut.	5		763	Tyr⇒Cys							Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		LBD			2650	TAC⇒TGC											
0158	PAIS	Substitut.	5	*	763	Tyr⇒Cys	12		normal	high		*	PolyGln tract short (only 12 repeats)	Male	Ambiguous	pos	McPhaul et al;J Clin Inv 87:1413,1991:Batch&al
		LBD			2650	TAC⇒TGC											
0850	CAIS	Substitut.	5		763	Tyr⇒Stop							Diag at 1 wk-Bilateral gonadectomy at 3wks	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2651	TAC⇒											
0160	CAIS	Substitut.	5	*	764	Phe⇒Leu			low		high			Female	Normal	neg	Marcelli et al; J clin Invest 94: 1642-1650, 1994
		LBD			2652	TTC⇒											
0161	CAIS	Substitut.	5		764	Phe⇒Leu			zero					Female	Normal		Ris-Stalpers et al: Pediatric Res,36; 227 -234, 1994
		LBD			2652	TTC⇒CTC											
0162	CAIS	Substitut.	5		764	Phe⇒Leu			low	normal				Female	Normal		Pinsky et al; Clin Inv Med, 15:456, 1992
		LBD			2654	TTC⇒TTG											
0163	CAIS	Substitut.	5	*	*	765	Ala⇒Thr			zero				Female	Normal		Bevan et al; J Steroid Biochem Molec. Biol 61: 19-26, 1997
		LBD				2655	GCC⇒ACC										
0164	CAIS	Substitut.	5		*	765	Ala⇒Thr			zero				Female	Normal		Merkabi et al;75th US Endo Soc Meeting Abstr 602, 1993
		LBD				2655	GCC⇒ACC										
0165	CAIS	Substitut.	5		*	765	Ala⇒Thr							Female	Normal		Sweet et al; Fertil Sterilty 58: 703, 1992
		LBD				2655	GCC⇒ACC										
0166	CAIS	Substitut.	5		*	765	Ala⇒Thr			zero				Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
		LBD				2655	GCC⇒ACC										
0311	CAIS	Substitut.	5		*	765	Ala⇒Thr	27						Female	Normal		Ko et al; J Reprod. Med 42: 424- 427, 1997
		LBD				2655	GCC⇒ACC										
0382	CAIS	Substitut.	5		*	765	Ala⇒Thr							Female	Normal		Giwercman et al; Human Genetics 103: 529-531, 1998
		LBD				2655	GCC⇒ACC										
0454	CAIS	Substitut.	5		*	765	Ala⇒Thr							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		LBD				2655	GCC⇒ACC										
0455	CAIS	Substitut.	5		*	765	Ala⇒Thr							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		LBD				2655	GCC⇒ACC										
0456	CAIS	Substitut.	5		*	765	Ala⇒Thr						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		LBD				2655	GCC⇒ACC										
0585	CAIS	Substitut.	5			765	Ala⇒Thr						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
		LBD				2655	GCC⇒ACC										
0586	PAIS	Substitut.	5			765	Ala⇒Thr						Reduced immunoreactive AR	Female	Ambiguous		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
		LBD				2655	GCC⇒ACC										
0730	CAIS	Substitut.	5			765	Ala⇒Thr							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
		LBD				2655	GCC⇒ACC										
0520	PAIS	Substitut.	5			765	Ala⇒Ser										Chavez et al; Clin Genet 59:: 185-188, 2001
		LBD				2655	GCC⇒TCC										
0167	CAIS	Substitut.	5			765	Ala⇒Val	20		zero				Female	Normal		Pinsky et al, Clin Inv Med, 15:456, 1992
		LBD				2656	GCC⇒GTC										
0665	PAIS	Substitut.	5			766	Pro⇒Ser						Somatic mosaicism	Female	Ambiguous		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
		LBD				2658	CCT⇒TCT										
0168	CAIS	Substitut.	5	*		766	Pro⇒Ser			low	high	high		Female	Normal	pos	Marcelli et al; J Clin Invest 94: 1642-1650. 1994
		LBD				2658	CCT⇒TCT										
0457	CAIS	Substitut.	5			766	Pro⇒Ser							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		LBD				2658	CCT⇒TCT										
0681	CAIS	Substitut.	5			766	Pro⇒Ala						bilateral inguinal hernia	Female	Normal		Bouvattier et al; J Clin Endocrinol & Metab 87: 29-32, 2002
		LBD				2658	CCT⇒GCT										

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0543 CAIS	Substitut.	5 LBD		766 2658	Pro⇒Ala CCT⇒GCT			normal	high		2 affcted siblings variable phenotype	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0611 CAIS	Substitut.	5 LBD		766 2658	Pro⇒Ala CCT⇒GCT						Twin sisters	Female	Normal	pos	Correa et al; 16th Meet. Latin Amer.Soc Ped Endo. Abstr: 26, 2003
0587 CAIS	Deletion	5 LBD		766 2659	Pro⇒Leu CΔCT⇒CTG						1 nt. del.-frameshift & stop in codon 787 low immunoreact AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0169 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						Single nt. deletion causing frameshift & stop in Codon 807	Female	Normal	pos	Baldazzi et al; Hum Mol Genet 3:1169-1170, 1994
0388 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						Single nt. deletion causing frameshift & stop in Codon 807	Female	Normal		Chung et al; Molecules & Cells 8: 741-745, 1998
0458 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1nt. del-frameshit & stop in Codon 807 Woolfian remnants	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0459 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1nt. del-frameshit & stop in Codon 807 Woolfian remnants	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0561 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1nt. del frame shift & stop in Codon 807 in 2 unrelat individs	Female	Normal		Guillen et al; An Esp Pediatr 56: 341-352, 2002
0588 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1 nt. del framshift & stop Codon 807 no immunoreact AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0853 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1nt. del framshift & stop in Codon 807 Diag - 7yr Woolf rem	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0953 CAIS	Deletion	5 LBD		766 2660	Pro⇒Pro CCΔT⇒CCG						1nt. del framshift & stop in Codon 807 Male gender identity	Female	Normal		T'Sjoen et al. Arch Sex Behav 2010
0893 CAIS	Substitut.	5 LBD		767 2661	Asp⇒Tyr GAT⇒TAT						Bone size intermediate between male and female	Female	Normal		Taes et al. Bone 45: 392-397, 2009
0170 CAIS	Substitut.	5 LBD		767 2663	Asp⇒Glu GAT⇒GAG			v low				Female	Normal		Lobaccaro et al; Pediatr Res, 33.Abstr 115, 1993
0343 CAIS	Substitut.	5 LBD		767 2663	Asp⇒Glu GAT⇒GAG							Female	Normal		Melo et al; 80th US Endo Soc Meeting Abstr P2-44, 1998
0544 PAIS	Substitut.	5 LBD		768 2664	Leu⇒Met CTG⇒ATG			normal	high		2 affected siblings with variable phenotype	Female	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0637 CAIS	Substitut.	5 LBD		768 2664	Leu⇒Val CTG⇒GTG	20					Testis located in labia majora	Female	Normal		Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0460 CAIS	Substitut.	5 LBD		768 2665	Leu⇒Pro CTG⇒CCG							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0666 CAIS	Substitut.	5 LBD		770 2671	Phe⇒Stop TTC⇒TAA						Somatic mosaicism sister hereozygous mother som mosaic ?	Female	Normal	pos	Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
0171 PAIS	Substitut.	5 LBD		771 2673	Asn⇒His AAT⇒CAT							Female	Ambiguous		Hiort et al; Hum Mol Genet 3: 1163-1166 1994
0526 PAIS	Substitut.	5 LBD	*	771 2673	Asn⇒His AAT⇒CAT				high		Size & level of expression of AR normal	Female	Ambiguous		Zhu et al, 83rd US Endo Soc Meeting, Abstr P2-34, 2001
0172 CAIS	Substitut.	5 LBD		772 2676	Glu⇒Stop GAG⇒TAG			zero				Female	Normal		Imasaki et al; Endocrine Journal 42: 643-648 1995
0839 CAIS	Deletion	5 LBD		772 2676	Glu⇒0 ΔGAGT⇒						4 nt deletion leading to stop in codon 787	Female	Normal		Jeske et al; J Pediatr Endocrinol Metab 20:893-908, 2007
0173 PAIS	Substitut.	5 LBD		772 2677	Glu⇒Gly GAG⇒GGG			low	high						Tincello et al; Clinical Endocrinology 46: 497 -506, 1997
0174 PAIS	Substitut.	5 LBD	*	772 2677	Glu⇒Ala GAG⇒GCG	25	23	normal	normal	high		Male	Ambiguous		Shkolny et al; J Clin Endocrinol & Metab 84: 805-810, 1999
0863 CAIS	Substitut.	5 LBD	*	774 2682	Arg⇒Cys CGC⇒TCC						Prenat Diagosis bilateral gonadectomy at 5yrs	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain		CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0878	CAIS	Substitut.	5 LBD		*	774 2682	Arg⇒Cys CGC⇒TCC							Female	Normal	pos	Qin et al; Zhonghua Fu Chan Ke Za Za 43:828 -830, 2008
0986	CAIS	Substitut.	5 LBD		*	774 2682	Arg⇒Cys CGC⇒TCC	27	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0336	CAIS	Substitut.	6 LBD	*	*	774 2682	Arg⇒Cys CGC⇒TGC	26	23	normal	normal			Female	Normal		Prior et al; Am J Hum Genet, 51:143, 1992
0176	CAIS	Substitut.	6 LBD	*	*	774 2682	Arg⇒Cys CGC⇒TGC	27	19	zero				Female	Normal	pos	Prior et al; Am J Hum Genet, 51:143, 1992
0177	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		Mebarki et al; 72nd US Endo Soc Meeting, Abstr 791, 1990
0178	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC							Female	Normal		Hiort et al; J Pediatrics 132: 939-943, 1998
0179	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC			v low	high			Female	Normal	neg	Marcelli et al; J Clin Endocrinol & Metab 73: 318, 1991
0180	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC							Female	Normal		Jakubiczka et al; Human Mutation 9: 57 -61, 1997
0331	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC							Female	Normal		Komori et al; Arch Gynecol & Obstetrics 261: 95-100, 1998
0175	CAIS	Substitut.	6 LBD	*	*	774 2682	Arg⇒Cys CGC⇒TGC			v low				Female	Normal		Brown et al; Mol Endocrinol, 4:1759-72, 1990
0355	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC						mosaic-de novo mutation	Female	Normal	neg	Hiort et al; J Pediatrics 132: 939-943, 1998
0589	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC						Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0599	CAIS	Substitut.	6 LBD		*	774 2682	Arg⇒Cys CGC⇒TGC							Female	Normal		Scheiber et al; J Pedatric Endocrinol & Metab. 16: 367-373,
0731	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC						sister affected	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
0732	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC						mother heterozygous	Female	Normal	pos	Ledig et al: Horm Res 63:263-269, 2005
0182	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC			low	normal	*		Female	Normal		Batch et al; Hum Mol Genet, 1:497, 1992
0183	CAIS	Substitut.	6 LBD	*	*	774 2683	Arg⇒His CGC⇒CAC			v low	high			Female	Normal		DeBellis et al; Mol Endocrinol, 6:1909-20, 1992
0184	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC							Female	Normal		Hiort et al; Am J Med Genet. 63; 218-222, 1996
0461	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0462	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0181	CAIS	Substitut.	6 LBD	*	*	774 2683	Arg⇒His CGC⇒CAC			normal	high	high	* mutation found in two unrelated families	Female	Normal	pos	Prior et al; Am J Hum genet, 51:143, 1992
0987	CAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC	20	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0185	PAIS	Substitut.	6 LBD		*	774 2683	Arg⇒His CGC⇒CAC										Quigley et al; Endocrin Reviews 16: 271, 1995
0186	CAIS	Substitut.	6 LBD		*	779 2697	Arg⇒Trp CGG⇒TGG							Female	Normal		Hiort et al; Hum Mol Genet 3: 1163-1166 1994
0187	CAIS	Substitut.	6 LBD	*	*	779 2697	Arg⇒Trp CGG⇒TGG							Female	Normal		Morono et al; Human Mutation 6: 152-162, 1995

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0188	CAIS	Substitut.	*	779	Arg⇒Trp								Female	Normal		Sinnecker et al; Eur J. Pediatr. 156: 7-14, 1997
		6 LBD		2697	CGG⇒TGG											
0463	CAIS	Substitut.	*	779	Arg⇒Trp								Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		6 LBD		2697	CGG⇒TGG											
0621	CAIS	Substitut.	*	779	Arg⇒Trp	21		low				AR partially purified appears truncated (43 kD)	Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
		6 LBD		2697	CGG⇒TGG											
0638	CAIS	Substitut.	*	779	Arg⇒Trp	24						Testis located in inguinal region	Female	Normal	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
		6 LBD		2697	CGG⇒TGG											
0733	CAIS	Substitut.	*	779	Arg⇒Trp								Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
		6 LBD		2697	CGG⇒TGG											
0189	PAIS	Substitut.	*	780	Met⇒Ile			normal	high		*		Female	Ambiguous		Bevan et al; Hum Mol Genet, 5: 265-273, 1996
		6 LBD		2702	ATG⇒ATA											
0190	PAIS	Substitut.		780	Met⇒Ile	20	23	normal	high	high		1 family member - male. Rest of family - females	Female / Male	Ambiguous	pos	Pinsky et al; Clin Inv Med, 15:456, 1992
		6 LBD		2702	ATG⇒ATA											
0191	PAIS	Substitut.		780	Met⇒Ile											Brinkmann et al; J Steroid Biochem & Mol Biol 53: 443, 1995
		6 LBD		2702	ATG⇒ATA											
0192	PAIS	Substitut.		780	Met⇒Ile							A brother to mutation 0305	Male	Ambiguous	pos	Rodien et al; J Clin Endo & Metab 81: 2904 -2908, 1996
		6 LBD		2702	ATG⇒ATA											
0305	CAIS	Substitut.		780	Met⇒Ile							2 sisters to mutation 0192	Female	Normal	pos	Rodien et al; J Clin Endo & Metab 81: 2904 -2908, 1996
		6 LBD		2702	ATG⇒ATA											
0193	CAIS	Substitut.		780	Met⇒Ile								Female	Normal		Jakubiczka et al; Human Mutation 9: 57 -61, 1997
		6 LBD		2702	ATG⇒ATA											
0464	CAIS	Substitut.		780	Met⇒Ile			low	high			Epidiymis & Vas deferens present - Cousin 660	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		6 LBD		2702	ATG⇒ATA											
0660	CAIS	Substitut.		780	Met⇒Ile							Epididymis & vas deferens present -Cousin 464	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		6 LBD		2702	ATG⇒ATA											
0682	PAIS	Substitut.		780	Met⇒Ile									Ambiguous		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
		6 LBD		2702	ATG⇒ATA											
0743	PAIS	Substitut.		780	Met⇒Ile							2 affected siblings	Female	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
		6 LBD		2702	ATG⇒ATA											
0774	MAIS	Substitut.		780	Met⇒Ile							Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
		6 LBD		2702	ATG⇒ATA											
0988	CAIS	Substitut.		781	Tyr⇒Asp	20	18	low	high			Mother & sister heterozygotes, 2 aunts wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		6 LBD		2701	TAC⇒GAC											
0194	Prostate cancer	Substitut.		782	Ser⇒Asn							Somatic mutation	Male	Normal		Tilley et al; 2: Clinical Cancer Res. 2: 277-285, 1996
		6 LBD		2707	AGC⇒AAC											
0383	CAIS	Substitut.	*	784	Cys⇒Tyr			zero				No transactivation capacity	Female	Normal		Giwercman et al; Human Genetics 103: 529-531, 1998
		6 LBD		2713	TGT⇒TAT											
0864	CAIS	Substitut.		784	Cys⇒Tyr							Diagnosis & bilateral gonadectomy at 16yrs	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
		6 LBD		2713	TGT⇒TAT											
0667	CAIS	Deletion		785	Val⇒Pro							Somatic mosaicism 2nt del causing fs & stop at codon 827	Female	Normal		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
		6 LBD		2715	ΔGTC⇒CCG											
0989	CAIS	Deletion		785	Val⇒Pro	19	18	zero	zero			2nt del causing fs & stop at codon 827	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		6 LBD		2715	ΔGTC⇒CCG											
0195	CAIS	Substitut.	*	786	Arg⇒Stop			zero					Female	Normal		Pinsky et al; Clin Inv Med, 15:456, 1992
		6 LBD		2718	CGA⇒TGA											
0557	CAIS	Substitut.	*	786	Arg⇒Stop								Female	Normal		Ignaccack et al; J Appl Genet 43: 109-114, 2002
		6 LBD		2718	CGA⇒TGA											
0600	CAIS	Substitut.	*	786	Arg⇒Stop								Female	Normal		Scheiber et al; J Pedatric Endocr Metab 16:367-373, 2003
		6 LBD		2718	CGA⇒TGA											

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0892 Prostate Cancer	Substitut.	6 LBD	*	786 2718	Arg⇒Stop CGA⇒TGA							+ΔQ86, Q867X Bicalutamide treated	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0939 Prostate Cancer	Substitut.	6 LBD	*	786 2718	Arg⇒Stop CGA⇒TGA							Untreated with anti-androgens	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0196 CAIS	Substitut.	6 LBD	*	787 2721	Met⇒Val ATG⇒GTG			zero					Female	Normal	pos	Nakao et al; J Clin Endocrinol Metab, 74:1152, 1992
0615 CAIS	Substitut.	6 LBD		787 2721	Met⇒Val ATG⇒GTG							Uterus present	Female	Normal		Van et al. Eur J. Pediatr. 162: 781-784, 2003
0990 CAIS	Substitut.	6 LBD		787 2723	Met⇒Ile ATG⇒ATA	23	18						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0991 CAIS	Substitut.	6 LBD		787 2723	Met⇒Ile ATG⇒ATA	23	17					1 sister affected	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0992 CAIS	Substitut.	6 LBD		787 2723	Met⇒Ile ATG⇒ATT	26	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0406 MAIS	Substitut.	6 LBD		788 2726	Arg⇒Ser AGG⇒AGT	24	23	normal	normal	high	*	Gynocomastia -high pitched voice-decreased body hair	Male	Normal	pos	Lumbroso et al 81st. US Endo Soc Meetings Abstr. P3-288, 1999
0716 PAIS	Substitut.	6 LBD		788 2726	Arg⇒Ser AGG⇒AGT				high				Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0735 CAIS	Duplicat	6 LBD		788 2726	Arg⇒ AGG⇒							Arg (AGG) duplicated	Male	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0197 MAIS	Substitut.	6 LBD	*	790 2730	Leu⇒Phe CTC⇒TTC			normal	low		*		Male	Near-normal male		Tsukada et al: J Clin Endorinol Metab, 79:1202, 1994
0842 CAIS	Substitut.	6 LBD		790 2731	Leu⇒Pro CTC⇒CCC								Female	Normal		Raicu et al; Asian J Androl 10:687-91, 2008
0993 CAIS	Deletion	6 LBD		792 2737	Gln⇒Arg ΔCAA⇒CGA	20	17					Heterozygous sister 2nt del causing fs & stop in Codon 827	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0198 MAIS	Substitut.	6 LBD		793 2741	Glu⇒Asp GAG⇒GAC			normal	normal			Inconsistent increases in gynocomastia & infertility	Male	Normal		Pinsky et al; Clin Inv Med, 15:456, 1992
0397 Normal	Substitut.	6 LBD		793 2741	Glu⇒Asp GAG⇒GAC							Homsosexual individual	Male	Normal		Macke et al; Am J Human Genetics 53: 844-852, 1993
0199 CAIS	Substitut.	6 LBD		794 2743	Phe⇒Ser TTT⇒TCT			zero					Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0200 CAIS	Substitut.	6 LBD		794 2743	Phe⇒Ser TTT⇒TCT								Female	Normal		Jakubiczka et al Human Mutation 9: 57-61, 1997
0907 PAIS	Substitut.	6 LBD		795 2747	Gly⇒Gly GGA⇒GGG			low	high			Silent mutation	Male	Ambiguous		Appari et al; J Mol Med 87: 623-632, 2009
0201 CAIS	Substitut.	6 LBD	*	796 2750	Trp⇒Stop TGG⇒TGA			v low					Female	Normal		Marcelli at al; J Clin Invest 85: 1522, 1990
0954 PAIS	Substitut.	6 LBD	*	798 2754	Gln⇒Glu CAA⇒GAA							Virilization post-gonadectomy heteroz P450OR p.Y601C	Female	Ambiguous		Idkowiak et al. J Clin Endocrinol Metab 95:3418-3527, 2010
0202 PAIS	Substitut.	6 LBD	*	798 2754	Gln⇒Glu CAA⇒GAA			normal	normal		*		Female	Ambiguous		Bevan et al; Hum Mol Genet, 5: 265-273, 1996
0203 PAIS	Substitut.	6 LBD		798 2754	Gln⇒Glu CAA⇒GAA			normal	normal							Quigley et al; Endocrine Reviews 16: 271, 1995
0204 PAIS	Substitut.	6 LBD		798 2754	Gln⇒Glu CAA⇒GAA								Female	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0205 Prostate cancer	Substitut.	6 LBD		798 2754	Gln⇒Glu CAA⇒GAA							Also present in genomic DNA	Male	Normal		Evans et al; Prostate 28: 162-171, 1996
0399 Prostate cancer	Substitut.	6 LBD		798 2754	Gln⇒Glu CAA⇒GAA							Somatic mutation Stage 4 tumor	Male	Normal		Castagnaro et al; Verh. Dtsch. Ges. Path. 77; 119-123, 1993

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0340	MAIS	Substitut.	*	798 2754	Gln⇒Glu CAA⇒GAA			normal			Azospermia	Male	Normal		Hiort et al; J Clin Endocrinol & Metab 85: 2810-2815, 2000
0381	MAIS	Substitut.	*	798 2754	Gln⇒Glu CAA⇒GAA			normal			Azoospermia - defective transactivation	Male	Normal		Wang et al; J Clin Endocrinol & Metab 83:
0775	MAIS	Substitut.		798 2754	Gln⇒Glu CAA⇒GAA						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0648	CAIS	Substitut.		798 2754	Gln⇒Stop CAA⇒TAA						Abdominal tumor - Sertoli cell adenoma	Female	Normal		Ignacek et al: Gynecol Endorinol 19: 178-181, 2004
0764	Prostate cancer	Substitut.		799 2758	Ile⇒Thr ATC⇒ACC						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0596	PAIS	Substitut.		800 2761	Thr⇒Ile ACC⇒ATC						Variable expressity- four additional males with MAIS	Male	Ambiguous	pos	Galli-Tsinopoulou et al. J Pediatr Endocrinol Metab16:149-154, 2003
0542	CAIS	Deletion		800 2762	Thr⇒Thr ACΔC⇒ACC						Single nt. del causing frameshift & stop in codon 807 1 affect sib	Female	Normal	pos	Boehmer et al; J Clin Endocrinol & Metab 86: 4151-4160, 2001
0521	PAIS	Substitut.		802 2767	Gln⇒Arg CAG⇒CGG										Chavez et al; Clin Genet 59:: 185-188, 2001
0498	CAIS	Substitut.		803 2769	Glu⇒Lys GAA⇒AAA			zero	zero			Female	Normal	pos	Sawai et al. J Hum Genet 45: 342-345, 2000
0563	CAIS	Substitut.		804 2772	Phe⇒Leu TTC⇒CTC						*de novo* mutation. + glaucoma & hyper-trophic pyloric stenos	Female	Normal	neg	Gad et al. Clin Genet. 63: 59-63, 2003
0865	CAIS	Substitut.		804 2772	Phe⇒Ile TTC⇒ATC						Diagnosis at 1mo	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0206	PAIS	Substitut.		806 2779	Cys⇒Tyr TGC⇒TAC										Brown et al; Eur J Pediatr 152: (Suppl 2) S62, 1993
0207	CAIS	Substitut.	*	807 2781	Met⇒Val ATG⇒GTG			low				Female	Normal		Morono et al; Human Mutation 6: 152-162, 1995
0639	CAIS	Substitut.		807 2781	Met⇒Val ATG⇒GTG	30		low			Testis located in inguinal region	Female	Normal	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0428	PAIS	Substitut.	*	807 2782	Met⇒Thr ATG⇒ACG			low			Treatment with topical DHT restored male genital	Female	Ambiguous		Ong et al; Lancet 354: 1444-1445, 1999
0208	CAIS	Substitut.		807 2782	Met⇒Arg ATG⇒AGG			zero				Female	Normal		Adeyemo et al; Hum Mol Genet, 2:1809, 1993
0403	PAIS	Substitut.		812 2796	Leu⇒Phe CTC⇒TTC							Female	Normal		Yaegashi et al; Tohoku J of Exp Med 187: 263 -272, 1999
0799	CAIS	Substitut.	*	812 2797	Leu⇒Pro CTC⇒CCC			zero			Severely impaired AR transactivation	Female	Normal		Jaaskelainen et al. Hum Mutat 27:291, 2006
0209	PAIS	Substitut.		814 2803	Ser⇒Asn AGC⇒AAC	20		normal			Hormone binding specificity alterted.	Female	Ambiguous		Pinsky et al; Clin Inv Med, 15:456, 1992
0210	MAIS	Substitut.		814 2803	Ser⇒Asn AGC⇒AAC	20		normal			Hormone binding specificity altered	Male	Normal	pos	Pinsky et al; Clin Inv Med, 15:456, 1992
0685	PAIS	Substitut.		817 2811	Pro⇒Ala CCA⇒GCA			normal	high		Mother a carrier	Male	Ambiguous	pos	Lombrosos et al; J Mol Endocrinol 32: 679 -687, 2004
0811	CAIS	Substitut.		817 2812	Pro⇒Leu CCA⇒CTA						Two sisters with CAIS mother a carrier	Female	Normal	pos	Turek-Plewa et al. Hum Genet 119:361, 2006
0501	CAIS	Substitut.		819 2818	Asp⇒Gln GAT⇒GGT						Also del of G (aa2702) caussing frameshift &	Female	Normal		Choi et al; Arch Gynecol Obstet 263: 201-205, 2000
0211	CAIS	Substitut.	*	820 2821	Gly⇒Ala GGG⇒GCG			normal	high	*	Also Leu 257 Pro, enhances thermolability	Female	Ambiguous	neg	Tanaka et al; Gynecological Endo. 12: 75-82, 1998
0212	PAIS	Substitut.		821 2823	Leu⇒Val CTG⇒GTG	24	23	normal	normal						Pinsky et al; Clin Inv Med, 15:456, 1992

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0776	MAIS	Substitut.		821	Leu⇒Val						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
		7 LBD		2823	CTG⇒GTG										
0513	MAIS	Substitut.	*	824	Gln⇒Lys						Gynocomastia-normal fertility - related to 514	Male	Normal	pos	Giwercman et al; J Clin Endocrinol & Metab 85: 2253-2259, 2000
		7 LBD		2832	CAA⇒AAA										
0514	MAIS	Substitut.	*	824	Gln⇒Lys						Gynocomastia-normal fertility - related to 513	Male	Normal	pos	Giwercman et al; J Clin Endocrinol & Metab 85: 2253-2259, 2000
		7 LBD		2832	CAA⇒AAA										
0875	PAIS	Substitut.	*	826	Phe⇒Leu	22	17	normal			Increased N/C terminal interaction & TIF2 co-activation	Male	Ambiguous	pos	Wong et al; Mol Cell Endocrinol 292:69-78, 2008
		7 LBD		2840	TTC⇒TTA										
0537	CAIS	Substitut.		827	Phe⇒Val							Female	Normal		Chavez et al; J Hum Genet. 46: 560-565, 2001
		7 LBD		2841	TTT⇒GTT										
0622	CAIS	Insertion		829	Glu⇒0	18					2nt insertion csuding frameshit and stop in Codon 833	Female	Normal	pos	MacLean et al. Hum Mutat. 23:287, 2004
		7 LBD		2847	GAA⇒+CA										
0522	CAIS	Substitut.		830	Leu⇒Val							Female	Normal		Chavez et al; Clin Genet 59:: 185-188, 2001
		7 LBD		2850	CTT⇒GTT										
0995	CAIS	Substitut.	*	831	Arg⇒Stop	19	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		7 LBD		2853	CGA⇒TGA										
0213	CAIS	Substitut.	*	831	Arg⇒Stop			zero				Female	Normal	pos	DeBellis et al; Mol Endocrinol, 6:1909-20, 1992
		7 LBD		2853	CGA⇒TGA										
0214	CAIS	Substitut.	*	831	Arg⇒Stop			zero				Female	Normal		Tincello et al; J Endocrinol, 132 Suppl, Abstr 87, 1992
		7 LBD		2853	CGA⇒TGA										
0215	CAIS	Substitut.	*	831	Arg⇒Stop			zero				Female	Normal		Ris-Stalpers et al; 74th Endo Soc Meeting, 1992
		7 LBD		2853	CGA⇒TGA										
0384	CAIS	Substitut.	*	831	Arg⇒Stop							Female	Normal		Giwercman et al; Human Genetics 103: 529-531, 1998
		7 LBD		2853	CGA⇒TGA										
0465	CAIS	Substitut.	*	831	Arg⇒Stop						Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		7 LBD		2853	CGA⇒TGA										
0500	CAIS	Substitut.	*	831	Arg⇒Stop							Female	Normal		Choi et al; Arch Gynecol Obstet 263: 201-205, 2000
		7 LBD		2853	CGA⇒TGA										
0515	CAIS	Substitut.	*	831	Arg⇒Stop						Harmatoma found in pubertal patient	Female	Normal		Chen et al; Fertilty & Sterility 74: 182-183, 2000
		7 LBD		2853	CGA⇒TGA										
0466	CAIS	Substitut.	*	831	Arg⇒Gln							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		7 LBD		2854	CGA⇒CAA										
0499	CAIS	Substitut.	*	831	Arg⇒Gln							Female	Normal		Choi et al; Arch Gynecol Obstet 263: 201-205, 2000
		7 LBD		2854	CGA⇒CAA										
0814	CAIS	Substitut.	*	831	Arg⇒Gln						"hamatomatous testes" present	Female	Normal		Goulis et al. Hirmines (Athens) 5: 200-204, 2006
		7 LBD		2854	CGA⇒CAA										
0216	CAIS	Substitut.	* *	831	Arg⇒Gln			v low				Female	Normal	pos	Brown et al; Mol Endocrinol, 4:1759-72, 1990
		7 LBD		2854	CGA⇒CAA										
0217	CAIS	Substitut.	*	831	Arg⇒Gln			zero			Found in two unrelated families	Female	Normal		McPhaul et al; J Clin Inv, 90: 2097, 1992
		7 LBD		2854	CGA⇒CAA										
0404	CAIS	Substitut.		831	Arg⇒Gln			zero				Female	Normal		Yaegashi et al; Tohoku J of Exp Med 187: 263 -272, 1999
		7 LBD		2854	CGA⇒CAA										
0524	CAIS	Substitut.		831	Arg⇒Gln			zero			Sertoli cell carcinoma	Female	Normal		Ko et al. Int. J. Gynocol. Pathol. 20: 196-199, 2001
		7 LBD		2854	CGA⇒CAA										
0994	CAIS	Substitut.		831	Arg⇒Gln	23	17				Mother heterozygous carrier	Female	Normal		Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		7 LBD		2854	CGA⇒CAA										
0218	CAIS	Substitut.	*	831	Arg⇒Leu	21	19	zero				Female	Normal		Shkolny et al; Human Mol Genetics 4: 515 -521,1995
		7 LBD		2854	CGA⇒CTA										
0307	CAIS	Substitut.	*	831	Arg⇒Leu	26	16	zero				Female	Normal		Shkolny et al; Human Mol Genetics 4: 515 -521,1995
		7 LBD		2854	CGA⇒CTA										

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot		Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0590	CAIS	Substitut.	7 LBD	*	831 2854	Arg⇒Leu CGA⇒CTA							Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0996	CAIS	Deletion			833 2859-61	Asn⇒0 ΔAAC⇒0	19	18						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0219	CAIS	Substitut.	7 LBD		834 2863	Tyr⇒Cys TAC⇒TGC			zero					Female	Normal		Wilson et al; J Clin Endocrinol Metab, 75:1474-8, 1992
0997	CAIS	Substitut.	7 LBD		838 2874	Leu⇒Val CTC⇒GTC	8	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0392	PAIS	Substitut.	7 LBD		838 2876	Leu⇒Leu CTC⇒CTT							Hypospadia and cryptorchidism - silent mutation	Male	Ambiguous		Nordenskjold et al Urological Res, 27: 49 -55, 1999
0765	Prostate cancer	Substitut.	7 LBD		838 2876	Leu⇒Leu CTC⇒CTT							patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0415	PAIS	Substitut.	7 LBD		840 2880	Arg⇒Ser CGT⇒AGT	25						Testis located in inguinal region. same family as 0832	Male	Ambiguous		Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0626	PAIS	Substitut.	7 LBD		840 2880	Arg⇒Ser CGT⇒AGT							Diagnosis at 6.5 yr. Reassigned as male after testosterone	Female	Ambiguous		Mazen et al. J Endocrinol Invest. 27:57-60, 2004
0832	PAIS	Substitut.	7 LBD		840 2880	Arg⇒Ser CGT⇒AGT							Testis located in inguinal region. same family as 0415	Male	Ambiguous		Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
0220	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT	20	16	normal	high	norm	*		Male	Ambiguous	pos	Beitel et al; J Clin Inv, 94: 546-554 1994
0221	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			low	high		*	Found in two unrelated individuals.	Female			McPhaul et al; J Clin Inv, 90: 2097, 1992
0222	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			normal	high			Sibling of 0308	Female	Ambiguous	pos	Bevan et al; Hum Mol Genet, 5: 265-273, 1996
0308	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			normal	high			Sibling of 0222	Male	Ambiguous	pos	Bevan et al; Hum Mol Genet, 5: 265-273, 1996
0387	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT							Transcriptional activity only at high conc of androgen				Georget et al; J Clin Endocrinol & Metab 83: 3597-3603, 1998
0698	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			normal	low			Family member of 0699 & 0700 with phenotypic varaiation	Male	Ambiguous	pos	Wang et al; Biochem Biophs Res Comm 335: 335-342, 2005
0699	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			normal	low			Family member of 0698 & 0700 with phenotypic varaiation	Male	Ambiguous	pos	Wang et al; Biochem Biophs Res Comm 335: 335-342, 2005
0700	PAIS	Substitut.	7 LBD	* *	840 2880	Arg⇒Cys CGT⇒TGT			normal	low			Family member of 0698 & 0699 with , phenotypic varaiation	Male	Ambiguous	pos	Wang et al; Biochem Biophs Res Comm 335: 335-342, 2005
0734	PAIS	Substitut.	7 LBD	*	840 2880	Arg⇒Cys CGT⇒TGT							brother affected, sister heterozygous	Male	Ambiguous	pos	Ledig et al: Horm Res 63:263-269, 2005
0385	PAIS	Substitut.	7 LBD	*	840 2880	Arg⇒Gly CGT⇒GGT			low				Reduced transactivation				Giwercman et al; Human Genetics 103: 529-531, 1998
0818	PAIS	Substitut.	7 LBD	*	840 2881	Arg⇒His CGT⇒CAT								Male	Ambiguous		Yen et al. Acta Paediatr Taiwan 46: 101-105, 2005
0683	CAIS	Substitut.	7 LBD	*	840 2881	Arg⇒His CGT⇒CAT							bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0820	CAIS	Substitut.	7 LBD	*	840 2881	Arg⇒His CGT⇒CAT								Female	Normal		Wang et al. Yi Chuan Xue Bao 33:19-25, 2006
0337	PAIS	Substitut.	7 LBD	* *	840 2881	Arg⇒His CGT⇒CAT	19		normal	high	high	*		Female	Ambiguous	pos	Beitel et al; J Clin Inv, 94: 546-554 1994
0224	PAIS	Substitut.	7 LBD	* *	840 2881	Arg⇒His CGT⇒CAT	18	24	normal	high	high	*		Female	Ambiguous	pos	Beitel et al; J Clin Inv, 94: 546-554 1994
0225	PAIS	Substitut.	7 LBD	*	840 2881	Arg⇒His CGT⇒CAT				high			Found in two unrelated families	Female	Ambiguous	pos in 1 fam	Hiort et al; J Clin Endocrinol Metab, 77:262-266, 1993

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0226	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			zero								McPhaul et al; J Clin Inv, 90:2097, 1992
0227	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			normal	normal		*	In same fam. persons raised as males with ambiguous genitalia	Female	Ambiguous	pos	Imasaki et al; Eur J Endorinol, 130: 569 -574, 1994
0228	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			low								Lumbroso et al; Eur J Endorinol 130: 327, 1994
0229	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			low								Imai et al; Annals of Clinical Biochem, 32: 482-486, 1995
0230	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT											Ghirri & Brown; Pediatr Res 33: Abstr.95, 1993
0231	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			low	high	high						Marcelli et al; J Clin Invest 94: 1642-1650, 1994
0232	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			normal	high		*		Female	Ambiguous	pos	Weidemann et al; Clin Endocrinology 45: 733 -
0223	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT			low	high				Female	Ambiguous		De Bellis et al; J Clin Endrcrinol Metab, 78:513, 1994
0668	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT							Somatic mosaicism	Male	Ambiguous		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
1008	PAIS	Substitut. 7 LBD	*	840 2881	Arg⇒His CGT⇒CAT	23	11					1 affected niece	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0233	PAIS	Substitut. 7 LBD		841 2884	Ile⇒Ser ATC⇒AGC			low	high				Female	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0234	CAIS	Substitut. 7 LBD		842 2887	Ile⇒Thr ATT⇒ACT			normal	high				Female	Normal	pos	Hiort et al; J Clin Endocrinol Metab, 77:262-266, 1993
0235	PAIS	Substitut. 7 LBD	*	842 2887	Ile⇒Thr ATT⇒ACT			low	high		*		Male	Ambiguous	pos	Weidemann et al Clin Endocrinlogy 45: 733 - 739, 1996
0591	CAIS	Substitut. 7 LBD		842 2887	Ile⇒Asn ATT⇒AAT							Normal immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0736	PAIS	Substitut. 7 LBD		843 2890	Ala⇒Glu GCA⇒GAA							de novo mutation	Male	Ambiguous	neg	Ledig et al: Horm Res 63:263-269, 2005
0494	Prostate cancer	Substitut. 7 LBD		846 2898	Arg⇒Gly AGA⇒GGA							Somatic mutation	Male	Normal		Marcelli et al; Cancer Research 60: 944-949, 2000
0957	CAIS	Insertion 7 LBD		848 2906	Asn⇒Lys AAT⇒AAAT							1nt insert - frameshift & stop in Codon 879	Female	Normal	neg	Wu et al. Fertility & Sterility 93:2076, e1-4, 2010
0236	CAIS	Insertion 7 LBD		848 2906	Asn⇒Lys AAT⇒AAAT			zero				1nt insert frameshift& stop in Codon 879& loss of 44 AA's	Female	Normal		Brinkmann et al; J Steroid Biochem Mol Biol 53: 443, 1995
0467	CAIS	Insertion 7 LBD		848 2906	Asn⇒Lys AAT⇒AAAT			zero				1nt insert causes frame-shift.	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0854	CAIS	7 LBD		848	Asn⇒ AAT⇒							Frameshit & stop in Codon 875 Diag & bilat gonadect at 7y	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0237	CAIS	Substitut. 7 LBD		853 2920	Ser⇒Stop TCA⇒TGA			zero					Female	Normal		Wilson et al; J Clin Endocrinol Metab, 75:1474-8, 1992
0238	CAIS	Substitut. 7 LBD		853 2920	Ser⇒Stop TCA⇒TGA			zero					Female	Normal		Jakubiczka et al; Human Mutation 9: 57 -61, 1997
0793	CAIS	Deletion 7 LBD		853 2922	Ser⇒ TCΔAA⇒							2 nt deletion	Female	Normal		Mueller et al. Hum Genet 119:673, 2006
0239	PAIS	Substitut. 7 LBD		854 2923	Arg⇒Lys AGA⇒AAA			low			*					McPhaul et al; J Clin Inv, 90:2097, 1992
0833	CAIS	Substitut. 7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC							gonads located in abdomen. Same family as 0604	Female	Normal		Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005

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0866 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC						Diagnosis at 3 wks	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0240 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		DeBellis et al; Mol Endocrinol 6:1909-20, 1992
0241 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC							Female	Normal		Tincello et al; J Endocrinol 132 Suppl, Abstr 87, 1992
0242 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		McPhaul et al; J Clin Inv, 90:2097, 1992
0243 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC							Female	Normal		Loboccaro et al; Pediat Res 33: Abstr 115, 1993
0244 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			low				Female	Normal	pos	Morono et al; Human Mutation 6: 152-162. 1995
0245 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		Sultan et al; J Steroid Biochem & Mol Biol:40
0246 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC							Female	Normal		Brinkmann et al; J Steroid Biochem & Mol Biol 53: 443, 1995
0247 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		Hiort et al; Am J Med Genet. 63: 218-222, 1996
0248 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			v low	high			Female	Normal	pos	Malmgren et al;Clin Genet. 50:202-205, 1996
0320 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC							Female	Normal		Komori et al: J Obstetrics & Gynocol. Res. 23: 277-81, 1997
0468 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			zero				Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0469 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			normal	high			Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0527 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC			v low	high			Female	Normal		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
0592 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC						v low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188, 2002
0640 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC	19		zero			Testis located in abdomen Same family as 0833	Female	Normal	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0737 CAIS	Substitut.	7 LBD	*	855 2925	Arg⇒Cys CGC⇒TGC						de novo mutation	Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0661 CAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC						Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0528 PAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC			normal	high	*		Male	Ambiguous		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
0684 CAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endorinol & Metab 87: 29-32, 2002
0688 CAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC						Mother a heterozyote	Female	Normal		Skordis et al; j pediatr Endocrinol Metab 18: 309-313, 2005
0745 PAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC						sibling of 0746	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
0746 PAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC			normal	high		niece of 0745 also Pro398Ser	Female	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
0777 MAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
0251 PAIS	Substitut.	7 LBD	*	855 2926	Arg⇒His CGC⇒CAC			normal	high						Chang et al; 73rd Endo Soc Meeting, Abstr 28, 1991

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0252	PAIS	Substitut.	7 LBD	*	*	855 2926	Arg⇒His CGC⇒CAC			normal	high		*	Servere hypospadia	Male	Ambiguous	pos	Batch et al; Hum Mol Genet, 1:497, 1992
0253	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC								Male	Ambiguous		Hiort et al; Am J Med Genet. 63: 218-222. 1996
0254	PAIS	Substitut.	7 LBD	*	*	855 2926	Arg⇒His CGC⇒CAC			zero					Female	Ambiguous	pos	Weidemann et al; Clin Endocrinology 45: 733 -
0255	PAIS	Substitut.	7 LBD	*	*	855 2926	Arg⇒His CGC⇒CAC			low	high	norm			Female	Ambiguous		Marcelli et al; J Clin Invest, 94:1642-1650, 1994
0301	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC	14						Brother of 0302 somatic & germ-line muts. in mother	Male	Ambiguous	pos	Boehmer et al; Am J Hum Genetics 60: 1003 -6, 1997
0250	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC			zero					Female	Ambiguous		Weidemann et al; Clin Endorinology 45: 733 -739, 1996
0302	PAIS	Substitut.	7 LBD	*	*	855 2926	Arg⇒His CGC⇒CAC	14						Sister of 0301. somatic & germ-line muts. in mother	Female	Ambiguous	pos	Boehmer et al; Am J Hum Genetics 60: 1003 -6, 1997
0249	CAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC			low					Female	Normal		McPhaul et al; J Clin Invest. 90: 2097, 1992
0344	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC	28		zero to normal				Sex changed at 1yr Male to female - Same family as 0834	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0834	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC							Same family as 0344	Male	Ambiguous	pos	Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
0998	CAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC	20	14					1 affected aunt; mother & 2 aunts heterozygotes	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
1009	PAIS	Substitut.	7 LBD		*	855 2926	Arg⇒His CGC⇒CAC	18	17					1 affected sister, mot & 2 aunts heterozygote carriers	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0641	PAIS	Substitut.	7 LBD			856 2930	Arg⇒His CGC⇒CAC	19		zero to normal				Testis located in inguinal region	Male	Ambiguous	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0716	PAIS	Substitut.	7 LBD			856 2930	Arg⇒His CGC⇒CAC				high				Male	Ambiguous		Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
0470	CAIS	Substitut.	7 LBD			856 2930	Phe⇒Leu TTC⇒TTG								Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0662	CAIS	Substitut.	7 LBD			856 2930	Phe⇒Leu TTC⇒TTG							Also Ser865Pro-twin of 0663	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0663	CAIS	Substitut.	7 LBD			856 2930	Phe⇒Leu TTC⇒TTG							Also Ser865Pro-twin of 0662	Female	Normal	pos	Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0356	CAIS	Substitut.	7 LBD			857	Tyr⇒ Stop TAC⇒							de novo mutation	Female	Normal	neg	Hiort et al; J Pediatrics 132: 939-943, 1998
0941	CAIS	Insertion	7 LBD			857 2933	Tyr⇒ Stop TAC⇒ TAaC							1nt insertion resulted in stop codon Mother & sister carriers	Female	Normal	pos	Turek-Plewa et al. Hum Genet 125:34, 2009
0753	CAIS	Substitut.	7 LBD	*		859 2937	Leu⇒Phe CTC⇒TTC			v low				In silico analysis showed effect on ligand-binding pocket	Female	Normal		Rajender et al. J Andrology 28:772-6, 2007
0867	CAIS	Substitut.	7 LBD			860 2941	Thr⇒ Asn ACC⇒AAC							Diagnosis at birth bilateral gonadectomy at 15yrs	Female	Normal		Cheikhelard et al. J Urol 180:1496-1501, 2008
0256	CAIS	Substitut.	7 LBD			863 2950	Leu⇒Arg CTG⇒CGG								Female	Normal		Brown et al; Eur J Pediatr 152: (Suppl 2) S62, 1993
0257	CAIS	Substitut.	7 LBD	*		864 2952	Asp⇒ Asn GAC⇒AAC			low				Transactivation activity increases with horm.	Female	Normal		Bevan et al; J Steroid Biochem Molec. Biol 61: 19-26, 1997
0471	CAIS	Substitut.	7 LBD			864 2952	Asp⇒ Asn GAC⇒AAC							Epididymis & vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0472	CAIS	Substitut.	7 LBD			864 2953	Asp⇒ Gly GAC⇒GGC			zero				Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004

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0258	CAIS	Substitut.	*	864	Asp⇒Gly			zero				Female	Normal		DeBellis et al; Mol Endocrinol, 6:1909-20, 1992
		7 LBD		2953	GAC⇒GGC										
0486	CAIS	Substitut.		865	Ser⇒Pro							Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
		7 LBD		2955	TCC⇒CCC										
0560	CAIS	Substitut.		865	Ser⇒Pro						de novo mut. also Phe868Leu mut -no effect horm binding	Female	Normal	pos	Mongan et al; J Clin endocrinol Metab 87: 1057-1061, 2002
		7 LBD		2955	TCC⇒CCC										
0855	CAIS			866	Val⇒						Frameshit & stop in Codon 875 Diag 15yr Bilat gonadect at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		7 LBD		2955	GTG⇒										
0856	CAIS			866	Val⇒						Frameshit & stop in Codon 875 Diag & Bilat gonadect at 11yr	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		7 LBD		2955	GTG⇒										
0259	PAIS	Substitut.		866	Val⇒Leu	21		normal	high			Male	Ambiguous	pos	Saunders et al; Clin Endocrinol 37:214, 1992
		7 LBD		2958	GTG⇒TTG										
0345	PAIS	Substitut.		866	Val⇒Leu	25		normal	high			Male	Ambiguous		Saunders et al; Clin Endocrinol, 37:214, 1992
		7 LBD		2958	GTG⇒TTG										
0260	PAIS	Substitut.	*	866	Val⇒Leu			normal	high			Male	Ambiguous	pos	Kazemi-Esfarjani et al; Mol Endocrinol, 7:37 -46, 1993
		7 LBD		2958	GTG⇒TTG										
0261	PAIS	Substitut.		866	Val⇒Leu				high			Male	Ambiguous	pos	Hiort et al; J Clin Endocrinol Metab, 77:262-266, 1993
		7 LBD		2958	GTG⇒TTG										
0262	PAIS	Substitut.	*	866	Val⇒Leu			zero							Merkabi et al; 75th US Endo Soc Meeting Abstr 602, 1993
		7 LBD		2958	GTG⇒										
0263	CAIS	Substitut.	*	* 866	Val⇒Met	20	16	normal	high			Female	Normal		Kazemi-Esfarjani et al; Mol Endocrinol, 7:37 -46, 1993
		7 LBD		2958	GTG⇒ATG										
0264	CAIS	Substitut.	*	* 866	Val⇒Met			normal	high			Female	Normal		Weidemann et al; Clin Endocrinology 45: 733 -
		7 LBD		2958	GTG⇒ATG										
0265	CAIS	Substitut.		* 866	Val⇒Met			normal	high	*		Female	Normal		Lubahn et al; Proc Natl Acad Sci. 86: 9534, 1989
		7 LBD		2958	GTG⇒ATG										
0266	PAIS	Substitut.		* 866	Val⇒Met					*					McPhaul et al; J Clin Inv, 90:2097, 1992
		7 LBD		2958	GTG⇒ATG										
0267	PAIS	Substitut.		* 866	Val⇒Met				high	*	de novo mutation - mosaic 2 functionally diff AR's	Female	Ambiguous	neg	Hiort et al; J Pediatrics 132: 939- 943, 1998
		7 LBD		2958	GTG⇒ATG										
0373	Prostate cancer	Substitut.		* 866	Val⇒Met						Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
		7 LBD		2958	GTG⇒ATG										
0473	CAIS	Substitut.		* 866	Val⇒Met						Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		7 LBD		2958	GTG⇒ATG										
0474	CAIS	Substitut.		* 866	Val⇒Met			zero			Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		7 LBD		2958	GTG⇒ATG										
0475	CAIS	Substitut.		* 866	Val⇒Met			zero			Epididymis present	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
		7 LBD		2958	GTG⇒ATG										
0607	CAIS	Substitut.		* 866	Val⇒Met			zero				Female	Normal		Holterhaus et al; Genome Biology 4: R37
		7 LBD		2958	GTG⇒ATG										
0738	PAIS	Substitut.		* 866	Val⇒Met							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
		7 LBD		2958	GTG⇒ATG										
0739	CAIS	Substitut.		* 866	Val⇒Met							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
		7 LBD		2958	GTG⇒ATG										
0574	Prostate Cancer	Substitut.		* 866	Val⇒Met						Somatic mutation. + orchiectomy treatment Horm-	Male	Normal		Hyytinen et al; Lab Invest. 82: 1591-1598, 2002
		7 LBD		2958	GTG⇒ATG										
0778	MAIS	Substitut.		* 866	Val⇒Met						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65:606-610, 2006
		7 LBD		2958	GTG⇒ATG										
0268	CAIS	Substitut.		866	Val⇒Glu							Female	Normal		McPhaul et al; J Clin Inv, 90:2097, 1992
		7 LBD		2959	GTG⇒GAG										

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot		Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0851	CAIS	Substitut.	7		867	Gln⇒ Stop							Diag at 2.5 yr	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2961	CAG⇒ TAG											
0852	CAIS	Substitut.	7		867	Gln⇒ Stop							Diag at 2 wk	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2961	CAG⇒ TAG											
0940	Prostate Cancer	Substitut.	7		867	Gln⇒ Stop							Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
		LBD			2961	CAG⇒ TAG											
0601	CAIS	Insertion	8		868	Pro⇒ Met							3 base insetion (ATG) at residue 868	Female	Normal		Scheiber et al; J Pedatric Endocrinol & Metab. 16: 367-373,
		LBD			2964	CCT⇒ ATG											
0269	PAIS	Substitut.	8	*	869	Ile⇒ Met			normal	high		*	Hypospadia	Male	Ambiguous	pos	Bevan et al; Hum Mol Genet 5: 265-273, 1996
		LBD			2969	ATT⇒ ATG											
0718	PAIS	Substitut.	8		869	Ile⇒ Met				high				Male	Ambiguous	pos	Deeb et al; Clinical Endocrinology 63: 56 -62, 2005
		LBD			2969	ATT⇒ ATG											
0951	PAIS	Substitut.	8	*	870	Ala⇒ Val							Micropenis only- Hetrozygous for SRD5A2 V89L	Male	Ambiguous		Bhangoo et al. Asian J Androl 12:561-566. 2010q
		LBD			2971	GCG⇒ GTG											
1010	PAIS	Substitut.	8	*	870	Ala⇒ Val	25	11						Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
		LBD			2971	GCG⇒ GTG											
0270	PAIS	Substitut.	8	*	870	Ala⇒ Val							Found in two unrelated families	Male	Ambiguous		Hiort et al; Eur J Pediatr, 153:317, 1994
		LBD			2971	GCG⇒ GTG											
0315	PAIS	Substitut.	8		870	Ala⇒ Gly							Servere hypospadias	Male	Ambiguous		Albers et al; J of Pediatrics 131: 388 -392, 1997
		LBD			2971	GCG⇒ GGG											
0271	PAIS	Substitut.	8	*	870	Ala⇒ Gly							de novo mutation	Female	Ambiguous	neg	Hiort et al; J Pediatrics 132: 939- 943, 1998
		LBD			2971	GCG⇒ GGG											
0562	MAIS	Substitut.	8	*	870	Ala⇒ Gly							bilateral gynecomastia normal fertility	Male	Normal		Zenteno et al; Horm Res 57: 90-93, 2002
		LBD			2971	GCG⇒ GGG											
0794	CAIS ?	Deletion	8		870	Ala⇒							1 nt. deletion				Mueller et al. Hum Genet 119:681, 2006
		LBD			2972	GCG⇒											
0272	MAIS	Substitut.	8	*	871	Arg⇒ Gly	26	24	normal	normal	norm		Gynocomastia & oligospermia	Male	Normal		Shkolny et al; J Clin Endocrinol & Metab 84: 805-810, 1999
		LBD			2973	AGA⇒ GGA											
0696	Prostate cancer	Substitut.	8	*	872	Glu⇒ Gln	24		normal	normal			Activated by estrodiol, progesterone & CPA	Male	Normal		Chen et al; The Prostate 63:395-406, 2005
		LBD			2976	GAG⇒ CAG											
0273	Prostate cancer	Substitut.	8		874	His⇒ Tyr							Som mut- stimulated by progesterone & oestrogen	Male	Normal		Taplin et al; New England J Med 332: 1393-1398, 1995
		LBD			2982	CAT⇒ TAT											
0274	Prostate cancer	Substitut.	8		874	His⇒ Tyr							Somatic mutation	Male	Normal		Tan et al; J of Urology 155: 340A, 1996
		LBD			2982	CAT⇒ TAT											
0538	CAIS	Substitut.	8		874	His⇒ Arg			zero					Female	Normal		Chavez et al; J Hum Genet. 46: 560-565, 2001
		LBD			2983	CAT⇒ CGT											
0868	CAIS	Substitut.	8		874	His⇒ Arg							Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0869	CAIS	Substitut.	8		874	His⇒ Arg							Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0870	CAIS	Substitut.	8		874	His⇒ Arg							Diagnosis at 2mo bilateral gnadectomy at 16yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0871	CAIS	Substitut.	8		874	His⇒ Arg							Diagnosis at 1yr bilateral gonadectomy at 15yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0872	CAIS	Substitut.	8		874	His⇒ Arg							Diagnosis at 7 yrs bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0873	CAIS	Substitut.	8		874	His⇒ Arg							Prenatal diagnosis & bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180:1496-1501, 2008
		LBD			2983	CAT⇒ CGT											
0275	LNCaP cell line	Substitut.	8		877	Thr⇒ Ala							Altered binding specificity - somatic mutation	Male	Normal		Veldscholte et al; Biochem Biophys Res Comm, 172:534, 1990
		LBD			2991	ACT⇒ GCT											

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0276	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						Somatic mutation 1/8 endocrine resistant therapy cases	Male	Normal		Suzuki et al; J Steroid Biochem Molec Biol 46:759, 1993
0277	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						6 out of 24 patients screened - somatic mutation	Male	Normal		Gaddipati et al; Cancer Res, 54: 2861-2864, 1994
0278	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						3 out of 22 cases in metastatic tissue - somatic mutation	Male	Normal		Suzuki et al; Prostate 29: 153-158, 1996
0279	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						Somatic mutation in bone metasteses of Prostate cancer	Male	Normal		Kleinerman et al; J of Urology 155: 624A, 1996
0432	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						Som mut found in 5 of 16 patients treated with flutamide	Male	Normal		Taplin et al; Cancer Research 59: 2511 -2515
0603	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						Somatic mutation - flutamide treated	Male	Normal		Taplin et al; J Clinical Oncology 21: 2673 -2678, 2003
0604	Prostate cancer	Substitut.	8 LBD	877 2991	Thr⇒Ala ACT⇒GCT						Somatic mutation - flutamide treated	Male	Normal		Taplin et al; J Clinical Oncology 21: 2673 -2678, 2003
0647	Prostate cancer	Substitut.	8 LBD *	877 2991	Thr⇒Ala ACT⇒GCT						+Gln640Stop- late stage disease- som mut -transactivates	Male	Normal		Ceraline et al; Intl J Cancer 108:152-157, 2003
0280	Prostate cancer	Substitut.	8 LBD *	877 2992	Thr⇒Ser ACT⇒AGT						Som mut. in 86% of isolates .Stimulated by estrogen &	Male	Normal		Taplin et al: New England J Med 332: 1393-1398, 1995
0539	PAIS	Substitut.	8 LBD	879 2997	Asp⇒Tyr GAC⇒TAC			normal				Male	Ambiguous		Chavez et al; J Hum Genet. 46: 560-565, 2001
0553	Prostate cancer	Substitut.	8 LBD	879 2998	Asp⇒Gly GAC⇒GCC						Treated with bicalumatide - somatic mutatation	Male	Normal		Taplin et al; J Clinical Oncology 21: 2673 -2678, 2003
0281	CAIS	Substitut.	8 LBD	881 3003	Leu⇒Val CTA⇒GTA						Somatic instabilty in polyglutamine tract	Female	Normal	pos	Davies et al; Clinical Endocrinology 43: 69 -77, 1995
0829	CAIS	Substitut.	8 LBD	881 3004	Leu⇒Pro CTA⇒GCA							Female	Normal	neg	Galani et al; Fertility & Sterility 2008
0282	CAIS	Substitut.	8 LBD	883 3009	Lys⇒Stop AAG⇒TAG			zero				Female	Normal	pos	Trifiro et al; Am J Med Genet, 40:493, 1991
0623	CAIS	Substitut.	8 LBD	884 3013	Ser⇒Stop TCA⇒TGA	23		v low				Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
0283	MAIS	Substitut.	8 LBD *	886 3018	Met⇒Val ATG⇒GTG	23	23	normal	normal	norm	Oligospermia-50% red. in transactivation	Male	Normal		Ghadessy et al. J. Clin. Endocrinol. 103:1517 -1525, 1999
0309	MAIS	Substitut.	8 LBD *	886 3018	Met⇒Val ATG⇒GTG	21	24	normal	normal	norm	Oligospermia-50% red. in transactivation	Male	Normal		Ghadessy et al. J. Clin. Endocrinol. 103:1517 -1525, 1999
0803	Liver cancer	Substitut.	8 LBD *	886 3018	Met⇒Val ATG⇒GTG	21	24				Som mut ? + T1034G in 5'UTR. found in tum & non-tumorus	Male	Normal		Yeh et al. Int J Cancer 120:1610-1617, 2007
0697	Prostate cancer	Substitut.	8 LBD *	886 3020	Met⇒Ile ATG⇒ATA	21		normal	normal		Increased response to TIF-2 & CBP w DHT w N-CoR > inhibition	Male	Normal		Chen et al; The Prostate 63:395-406, 2005
0593	PAIS	Substitut.	8 LBD *	888 3026	Ser⇒Lys AGC⇒AAG						no immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87; 182-188. 2002
0533	PAIS	Substitut. / Splice	8 LBD *	888 3026	Ser⇒Ser AGC⇒AGT	21	24	v low	normal		silent mut.-part exon 8+ part of 3' untransl also small amt. wt AR	Male	Ambiguous		Hellwinkel et al. J Clin Endocrinol & Metab 86: 2569-2575, 2001
0540	PAIS	Substitut/ Splice	8 LBD *	888 3026	Ser⇒Ser AGC⇒AGT			normal			silent mutation	Male	Ambiguous		Chavez et al. JHum Genet. 46: 560-565 2001
1011	PAIS	Substitut.	8 LBD *	888 3026	Ser⇒Ser AGC⇒AGT	23	17	zero	zero		mother heterozygote carrier	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
1012	PAIS	Substitut.	8 LBD *	888 3026	Ser⇒Ser AGC⇒AGT	25	18					Female	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0284	CAIS	Substitut.	8 LBD *	889 3027	Val⇒Met GTG⇒ATG			zero				Female	Normal		Pinsky et al; Clin Inv Med, 15:456, 1992

Accession # / Phenotype	Mutation type	Exon / Domain	Pathogenicity proven	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Androgen Binding: Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0285	PAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG			low	normal			Female	Ambiguous		De Bellis et al; J Clin Endocrinol Metab, 78:513, 1994
0321	PAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG							Female	Normal		Essawi et al; Disease Markers 13: 99-105, 1997
0476	CAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG			low	normal			Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0624	CAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG	22		low				Female	Normal		MacLean et al. Hum Mutat. 23:287, 2004
0740	CAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG							Female	Normal	neg	Ledig et al: Horm Res 63:263-269, 2005
0999	CAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ ATG	21	17				1 sister affected; mother heterozygote; 2 aunts wt	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
1013	PAIS	Substitut.	8 LBD	*	889 3027	Val⇒ Met GTG⇒ CTG	18	14					Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0433	Prostate cancer	Substitut.	8 LBD	*	890 3030	Asp⇒ Asn GAC⇒ AAC						Mutation also found in peripheral blood	Male	Normal		Taplin et al; Cancer Research 59:2511 -2515, 1999
0389	CAIS	Substitut.	8 LBD	*	892 3036	Pro⇒ Ser CCG⇒ TCG	26		low	high		Reduced transactivation	Female	Normal	neg	Peters et al; Mol & Cellular Endocrinol. 148: 47-53, 1999
0375	CAIS	Substitut.	8 LBD		892 3037	Pro⇒ Leu CCG⇒ CTG						Mutation found in two siblings	Female	Normal	pos	Knoke et al; Human Mutation 12: 220, 1998
0413	CAIS	Substitut.	8 LBD		892 3037	Pro⇒ Leu CCG⇒ CTG							Female	Normal		Kanayama et al; Int J Urology 6: 327-330, 1999
0741	CAIS	Substitut.	8 LBD		892 3037	Pro⇒ Leu CCG⇒ CTG							Female	Normal		Ledig et al: Horm Res 63:263-269, 2005
0742	CAIS	Substitut.	8 LBD		892 3037	Pro⇒ Leu CCG⇒ CTG							Female	Normal		Ledig et al: Horm Res 63:263-269, 2005
0795	CAIS	Deletion	8 LBD		894 3042	Met⇒ 0 ΔATG⇒						3 nt. deletion	Female	Normal		Mueller et al. Hum Genet 119:681, 2006
0766	Prostate cancer	Substitut.	8 LBD		895 3045	Met⇒ Val ATG⇒ GTG						patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98:1320-1325, 2006
0669	CAIS	Substitut.	8 LBD		895 3046	Met⇒ Thr ATG⇒ ACG						Somatic mosaicism	Male	Ambiguous		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
0386	CAIS	Substitut.	8 LBD	*	895 3046	Met⇒ Thr ATG⇒ ACG			low			Reduced transactivation	Female	Normal		Giwercman et al; Human Genetics 103: 529-531, 1998
1000	CAIS	Substitut.	8 LBD		895 3046	Met⇒ Thr ATG⇒ ACG	26	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0642	PAIS	Substitut.	8 LBD		898 3054	Ile⇒ Phe ATC⇒ TTC	19					Testis located in labia majora- Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0643	PAIS	Substitut.	8 LBD		898 3054	Ile⇒ Phe ATC⇒ TTC	19					Testis located in inguinal -Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0644	PAIS	Substitut.	8 LBD		898 3054	Ile⇒ Phe ATC⇒ TTC	19					Testis located in inguinal- Single vaginal & urethra opn	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0645	PAIS	Substitut.	8 LBD		898 3054	Ile⇒ Phe ATC⇒ TTC	19					Testis located in inguinal -Separated vaginal & urethra opn	Female	Ambiguous	pos	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
0286	CAIS	Substitut.	8 LBD		898 3055	Ile⇒ Thr ATC⇒ ACC						de novo mutation	Female	Normal	neg	Hiort et al; J Pediatrics 132: 939- 943, 1998
0649	PAIS	Substitut.	8 LBD	*	902 3065	Gln⇒ Lys CAA⇒ AAA				high	high	Involved in TIF2 N/C interaction	Male	Ambiguous	pos	Umar et al; J Cin Endocrinol & Metab 90:507-515, 2005
1014	PAIS	Substitut.	8 LBD		902 3065	Gln⇒ Lys CAA⇒ AAA	21	17				mother heterozygote carrier	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Poly Gly #	Androgen Binding: Bmax	Kd	Thermolabile k		Comments	Sex of rearing	External Genitalia	Family history	Reference
0287 Prostate cancer	Substitut.	8 LBD		902 3066	Gln⇒Arg CAA⇒CGA							Somatic mutation in 37% of isolates in initial cloning	Male	Normal		Taplin et al; New England J Med 332: 1393-1398, 1995
0288 PAIS	Substitut.	8 LBD		903 3069	Val⇒Met GTG⇒ATG			low				Qualitative binding abnormality				McPhaul et al; J Clin Inv, 90:2097, 1992
1001 CAIS	Substitut.	8 LBD		903 3069	Val⇒Leu GTG⇒TTG	23	18					Mother heterozygote carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0289 CAIS	Substitut.	8 LBD		904 3072	Pro⇒Ser CCC⇒TCC	27	23	normal	high				Female	Normal		Pinsky et al; Clin Inv Med, 15:456, 1992
0290 CAIS	Substitut.	8 LBD		904 3073	Pro⇒His CCC⇒CAC			zero					Female	Normal		McPhaul et al; J Clin Inv, 90:2097, 1992
0646 CAIS	Substitut.	8 LBD		904 3073	Pro⇒Val CCC⇒CGC	21						Testis located in inguinal region	Female	Normal	neg	Melo et al; J Clin Endorinol & Metab 88: 3241-3250, 2003
1002 CAIS	Deletion	8 LBD		905 3076	Lys⇒Arg AAAG⇒AGA	22	17					mother heterozygote carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0291 CAIS	Substitut.	8 LBD	*	907 3081	Leu⇒Phe CTT⇒TTT			low	normal			Decreased transactivat Epididymis & Vas	Female	Normal		Hannema et al. J Clin Endocrinol & Metab 89: 5815-5822, 2004
0292 PAIS	Substitut.	8 LBD	*	909 3087	Gly⇒Arg GGG⇒AGG			low	low			Also silent G to A mutation in codon 211	Female	Ambiguous	pos	Choong et al; J Clin Endocrinol Metab, 81: 236-243, 1996
0374 Prostate cancer	Substitut.	8 LBD		909 3088	Gly⇒Glu GGG⇒GAG							Somatic mutation	Male	Normal		Takahashi et al; Cancer Research 55: 1621 -1624, 1995
0327 Prostate cancer	Substitut.	8 LBD		910 3091	Lys⇒Arg AAA⇒AGA							Somatic mutation	Male	Normal		Watanabe et al; Jpn J Clin Oncol 27: 389 -393, 1997
0430 PAIS	Substitut.	8 LBD		911 3093	Val⇒Leu GTC⇒CTC	19						Servere oligozoospermia	Male	Ambiguous		Knoke et al; Andrologia 31: 199-201, 1999
0293 PAIS	Substitut.	8 LBD		913 3099	Pro⇒Ser CCC⇒TCC											Ghirri and Brown; Paed Res, 33(5) Suppl, Abstr 95, 1993
1015 PAIS	Substitut.	8 LBD		913 3099	Pro⇒Ser CCC⇒TCC			zero	zero				Female	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0958 CAIS	Substitut.	8 LBD		913 3100	Pro⇒Arg CCC⇒CGC								Female	Normal	neg	Wu et al. Fertility & Sterility 93:2076, e1-4, 2010
0844 CAIS	Deletion	8 LBD	*	915 3107	Tyr⇒Stop TAAT⇒TAG							12nt del.Stop in codon 915affect sister & aunts.neg N/C inter	Female	Normal	pos	Werner et al; Sex Dev 2:73-83, 2008
0318 CAIS	Substitut.	8 LBD	*	916 3110	Phe⇒Leu TTC⇒TTG			low	high		*		Female	Normal		Radnayr et al; J of Urology 158: 1553 -1556, 1997
0477 CAIS	Substitut.	8 LBD		917 3112	His⇒Arg CAC⇒CGC								Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0303 Prostate cancer	Substitut.	8 LBD	*	919 3118	Gln⇒Arg CAG⇒CGG							Somatic mutation	Male	Normal		Nazereth et al; 79th US Endo Soc Meetings Abstr. P2-489, 1997
0294 CAIS	Splice	exon1/ intron 1			⇒ gta⇒gtta	24	23					Insertion at +3 position of donor splice site	Female	Normal		Trifiro et al; Eur J Hum Genetics 5: 50-58, 1997
0906 CAIS	Splice	exon1/ intron 1			⇒ gtaag⇒gtaac							Substitution at +5 of donor spice site	Female	Normal	pos	Philibert et al; Fertility & Sterility 2009
0304 CAIS	Splice	exon2/ intron 2			⇒ ctg⇒cta							Substitution at +1 pos of donor splice site - lacks exon 2	Female	Normal	neg	Hellwinkel et al; J Steroid Biochem & Mol Biol 68: 1-9, 1999
0479 CAIS	Splice	exon2/ intron 2			⇒ ⇒			zero					Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0480 CAIS	Splice	exon2/ intron 2			⇒ ⇒								Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
1016 CAIS	Splice	exon2/ intron 2			⇒ cag⇒ccg	13	18					Substit at -2 of acceptor site. Also Lys590Glu mut	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010

Accession # / Phenotype	Mutation type	Pathogenicity proven Exon / Domain	CpG hot spot	Position Amino acid / Base	Change Amino acid / Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding Bmax	Kd	Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
1017	CAIS	Splice	exon2/ intron 2			⇒ cag⇒gag	26	18				Substit at -3 of acceptor site. Aborted fetus	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0295	CAIS	Splice	exon3/ intron 3			⇒ ggt⇒gat						Substitution at+1 position of donor splice site	Female	Normal		Evans et al; J Endocrinol 129 Suppl, Abstr 65, 1991
0478	CAIS	Splice	exon3/ intron 3			⇒ ggt⇒gat			normal	normal		Substitution at +1 position of donor splice site	Female	Normal		Ahmed et al; J Clin Endocrinol & Metab 85: 658-665, 2000
0296	CAIS	Splice	exon4/ intron 4			⇒ ggt⇒gtt			zero			+1 pos of donor site. Splice site activated & del of aa's 683-723	Female	Normal		Ris-Stalpers et al; Proc Natl AcadSci 87:7866 -70, 1990
0297	CAIS	Splice	exon6/ intron 6			⇒ gta⇒tta	21		zero			Substitution at +3 position of donor splice site	Female	Normal	pos	Pinsky et al; Eur J Hum Genetics 5: 50-58, 1997
0503	PAIS	Splice	exon6/ intron 6			⇒ taa⇒tat			low	normal		Substit. at +5 position of donor splice site, stop at + 79 bases	Female	Ambiguous		Sammarco et al; J Clin Endocrinol & Metab 85: 3256-3261, 2000
0541	CAIS	Splice	exon6/ intron 6			⇒aac aag⇒aac			zero			Substit. at +6 position of donor splice site.	Female	Normal		Chavez et al; J Hum Genet 46: 560-565, 2001
1018	CAIS	Splice	exon6/ intron 6			⇒ gta⇒ata	28	17	low	normal		Substit. at -44 pos of donor acceptor site	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95:1876-1888, 2010
0298	CAIS	Splice	exon7/ intron 7			⇒ tgt⇒tat			zero			Subst. at +1 pos of donor splice, - exon7, stop + 10 aa exon 8	Female	Normal	pos	Lim et al; Mol & Cell Endocrinology 131: 205 -210, 1997
0502	CAIS	Splice	exon7/ intron 7			⇒ tgt⇒tat						Substitution at + 1 position of donor splice site	Female	Normal		Choi et al; Arch Gynecol Obstet 263: 201-205, 2000
0670	CAIS	Splice	exon7/ intron 7			⇒ tgt⇒tat			normal	normal		Subst at + 1 pos of donor splice site somtaic mosaicsm	Female	Normal		Kohler et al; J Clin endocrinol & Metab 90: 106-111, 2005
0299	PAIS	Splice	intron 2/exon 3			⇒ gtt⇒gat						Subst. -11 pos accept. 2 transc; -exon3, +69 nt. 3 affct sib;var	Male	Normal	pos	Bruggenwirth et al; Am J Hum Genet 61: 1067-1077, 1997
0800	CAIS	Splice	intron 2/exon 3			⇒ gac⇒aac						Subst. -11 pos accept. 2 transc; -exon3, +69 nt.	Male	Normal		Jaaskelainen et al. Hum Mutat 27:291, 2006
0909	Prostate cancer	Splice	intron 2/exon 3			⇒ gac⇒aac						Subst. -11 pos accept. 2 transc; -exon3, +69 nt. found in 5/8 cases	Male	Normal		Steinkamp et al; Cancer Res 69:4434-4442, 2009
0809	Prostate cancer	Splice	intron 2/exon 3	*		⇒ gac⇒aac						+ 69 nt insertion = 23 aa affects AR intracellular traffic	Male	Normal		Jagla et al.; Endocrinology 148:4334-43, 2007
0317	Breast Cancer	Deletion				⇒ ⇒						-exon 3:higher express of mut.var in 7/13 breast cancer	Female	Normal		Zhu et al. Int J of Cancer 72:574-580, 1997
0594	CAIS	Substitut.	intron 2			ag⇒at ⇒							Female	Normal		Avila et al. J Clin Endocrinol Metab 87:182-188, 2002
0351	CAIS	Substitut.	intron 2			gt⇒at ⇒							Female	Normal		Hiort et al; J Pediatrics 132: 939- 943, 1998
0088	PAIS	Deletion	intron 2			⇒ ⇒			normal	normal		6 kb del at -18 pos of acceptor site 2 transcr: 1wt,1 - exon3	Male	Ambiguous	pos	Ris-Stalpers et al; Am J Hum Genet 54:609, 1994
0749	PAIS	Deletion	intron 2			⇒ ⇒			normal	high		6 kb del of intron 2 effects splicing	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
0750	PAIS	Deletion	intron 2			⇒ ⇒						related to 0750	Male	Ambiguous	pos	Boehmer et al; J Clin Endocrinol & Metab 86:4151-4160, 2001
0835	PAIS	Substitut.	intron 3			g⇒a ⇒						Male to female	Female	Ambiguous		Melo et al; Arq Bras Endocrinol Metab 49:87-97, 2005
1022	Premature ovarian failure	Substitut.	intron 3			tac⇒taa ⇒						Subst, at +9 of donor site. Patient had menopause at 18	Female	Normal		Panda et al. Gyncol Endocrinol 2010
0312	Prostate Cancer	Substitut.	5' UTR			**agc⇒atc** ⇒						+2 pos from transcription initiation site AR-TIS II	Male	Normal		Crociotto et al. J Urol 158:1599-1601, 1997
0313	Prostate cancer	5' UTR	5' UTR			**gcc⇒gac** ⇒						+214 pos from transcription initiation site AR-TIS II	Male	Normal		Crociotto et al. J Urol 158:1599-1601, 1997

Accession # / Phenotype	Mutation type	Pathogenicity proven / Exon / Domain	CpG hot spot	Position / Amino acid / Base	Change / Amino acid / Base	Exon 1 tracts: Poly Gln #	Exon 1 tracts: Poly Gly #	Androgen Binding: Bmax	Androgen Binding: Kd	Androgen Binding: Thermolabile k	Comments	Sex of rearing	External Genitalia	Family history	Reference
0323 Prostate cancer		3' UTR			⇒ ⇒						Som mut. olymorph seq 2820 -36 dwnstrm to transl init. site	Male	Normal		Paz et al. European Urology 31:209-215, 1997