

ANDROGEN RECEPTOR GENE MUTATIONS DATABASE

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Accession #	Phenotype	Mutation type	Pathogenicity proven Exon Domain	CpG hot spot	Position Amino acid HGVS nt Base HGVS base	Change Amino acid Base	Exon 1 tracts Poly Gln # Poly Gly #	Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
								Bmax	Kd	k					
0312	Prostate cancer	Substitut	5'UTR								+2 pos from transcription initiation site AR-TIS II (G>T)	Male	Normal		Crocioetto et al. J Urol 158: 1599-1601, 1997
1051	Breast Cancer	Substitut	5'UTR		-1101	105T >A					Mutation present in primary breast tumors from 2 patients	Female	Normal		Peter et al. BMC Cancer 12:132, 2012
0313	Prostate cancer	Substitut	5'UTR		-913	203C >A					+214 pos from transcription initiation site AR-TIS II	Male	Normal		Crocioetto et al. J Urol 158: 1599-1601, 1997
0754	PAIS	Substitut	1 Nterm	*	002 4	p.(Glu2 Lys) 1119G >A				high	20-50% reduction in mutant protein	Male	Ambiguous	pos	Choong et al. J Clin Invest. 98: 1423-1431, 1996
0960	CAIS	Substitut	1 Nterm	*	002 4	p.(Glu2 Lys) 1119G >A	27	13	low	high	Mother heterozygous carrier	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1059	CAIS	Substitut	1 Nterm		005 14	p.(Leu5 *) 1129T						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0751	CAIS	Deletion	1 Nterm		007 19	p.(Leu7 fs) 1134 del C					Elevated testosterone stop in codon 33 No WD development	Female	Normal	neg	Barbaro et al. Clin Endocrinol 66: 822-826, 2007
0963	CAIS	Duplicat	1 Nterm		015 39	p.(Pro15 fs) 1154_1157 dupGC CG	20	9			4 nt duplication (GCCG) causing fs and stop in codon 84	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1060	CAIS	Substitut	1 Nterm		020 58	p.(Arg20 *) 1173C >T						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
1061	CAIS	Substitut	1 Nterm		024 70	p.(Gln24 *) 1185C >T						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0624	CAIS	Deletion	1 Nterm		039 115	p.(Pro39 del) 1230_1232 del CCC					In frame deletion of 3 nt removing proline				Jung et al. Human Genetics 114: 222, 2004
0836	CAIS	Deletion	1 Nterm		040 118	p.(Arg40 fs) 1233 delA					1 nt del causing frameshift & stop in codon 174	Female	Normal	pos	Decaestecker et al. Fertility & Sterility 89: 1260 e3-7, 2008
0002	CAIS	Deletion	1 Nterm		042 125	p.(Pro42 fs) 1240 delC			zero		1 nt del causing frameshift & stop in Codon 174 1 aff sib	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0910	Prostate cancer	Substitut	1 Nterm		043 128	p.(Glu43 Gly) 1243A >G					Treated with Flutamide -occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0003	Prostate cancer	Substitut	1 Nterm		054 161	p.(Leu54 Ser) 1276T >C					Also Phe892Leu 3789T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0897	CAIS	Insertion	1 Nterm		055 163	p.(Leu55 fs) 1278_1279 insC						Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0808	Liver cancer	Substitut	1 Nterm		057 170	p.(Leu57 Gln) 1285T >A					Somatic mutation	Male	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0005	Prostate cancer	Substitut	1 Nterm		057 170	p.(Leu57 Gln) 1285T >A					Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0786	MAIS	Duplicat	1 Nterm		057	p.(Leu57 dup)					Leu duplicated - male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0004	Laryngeal cancer	Deletion	1 Nterm		057						30 nt deletion Somatic mutation	Male	Normal		Urushibata et al. 10th. Int. Cong. Endocrinol Abstr. P3-706, 1996
0815	Festicular cancer	Deletion	1 Nterm		057 169	p.(Leu57 del) 1284_1286 delC TG					3 nt deletion seminoma	Male	Normal		Garolla et al. Encdocrine Related Cancer 12: 645-655, 2005
1064	PAIS	Deletion	1 Nterm		057 169	p.(Leu57 del) 169_171 delC TG						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0411	Mental Retard.	Deletion	1 Nterm		058				8				normal normal	3 affected sibs - CAG = 8; normal maternal uncle CAG = 23	Male	Normal	pos	Kooy et al. Am J Med Genet 85: 389-393, 1999
1062	CAIS	Substitut	1 Nterm		058	172	p.(Gln58 *)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0612	MAIS ?	Substitut	1 Nterm		058	173	p.(Gln58 Leu)							2 out of 62 patients with male infertility	Male	Normal		Lund et al. Fertility & Sterility 79: 1647-148, 2003
0006	Kennedy Syndrome	Insertion	1 Nterm		058-080									Expansion of polyglutamine repeat	Male	Normal		LaSpada et al. Nature 352: 77-79, 1991
0007	Prostate cancer	Deletion	1 Nterm		058-080									Contraction of poly Gln repeats (24 to 18) Somatic mutation	Male	Normal		Schoenberg et al. Biochem Biophys Res Comm 198: 74-80 1994
0324	Prostate cancer	Deletion	1 Nterm		058-080									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	1 Nterm		058-080									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	1 Nterm		058-080									Contraction of poly Gln repeats (20 to 18) Somatic mutation	Male	Normal		Wallin et al. J Pathology 189: 559-653, 1999
0692	CAIS	Substitut	1 Nterm		059	176	p.(Gln59 *)					zero			Female	Normal		Holterhus et al. J Mol Med (Berl) 83: 1005-1013, 2005
0008	CAIS	Substitut	1 Nterm	*	060	178	p.(Gln60 *)				low	normal high		Normal upregulation.	Female	Normal	neg	Zoppi et al. J Clin Inv 19: 1105, 1993
0671	CAIS	Substitut	1 Nterm	*	060	178	p.(Gln60 *)							bilateral inguinal hernia.	Female	Normal		Bouvattier et al. J Clin Endocrinol Metab 87: 29-32, 2002
1063	CAIS	Substitut	1 Nterm		060	178	p.(Gln60 *)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0409	CAIS	Duplicat.	1 Nterm		060	179	p.(Gln60 fs)							either 1 nt insert or 2 nt del. - frameshift & stop in codon 83	Female	Normal		Zhu et al. J Clin Endocrinol Metab 84: 1590-1594, 1999
0009	Prostate cancer	Substitut	1 Nterm		064	188	p.(Gln64 Arg)							Also Leu831Pro 3607T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0846	CAIS	Substitut	1 Nterm		067	196	p.(Gln67 *)							Diag at 1Mo Bilateral gonadectomy at 18y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0894	CAIS	Substitut	1 Nterm		070	208	p.(Gln70 *)								Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0881	MAIS ?	Substitut	1 Nterm		070	209	p.(Gln70 Arg)			20,				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	217	p.(Gln73 *)							Somatic mosiac 2/3 mutant to 1/3mutant	Female	Normal		Mueller et al. Hum Genet 119: 681, 2006
0895	CAIS	Substitut	1 Nterm		076	226	p.(Gln76 *)								Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
0961	CAIS	Substitut	1 Nterm		076	226	p.(Gln76 *)						18	Truncated CAG repeat ?	Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0902	CAIS	Deletion	1 Nterm		076	227	p.(Gln76 del)								Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0807	Liver cancer	Substitut	1 Nterm		080	240	p.(Gln80 )							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		081	240	p.(Glu81 fs)			21	18			1 nt insertion causing frameshift and stop in codon 83	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0901	CAIS	Deletion	1 Nterm		082	244	p.(Thr82 fs)							5 base deletion	Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010
0965	CAIS	Deletion	1 Nterm		084		p.(Ser83 fs)			20	18	zero	zero	10 base deletion frameshift & stop in codon 171	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010



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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile						
0012	CAIS	Deletion	1 Nterm		142									Frameshift & deletion of codons 142-150 Stop in codon 156	Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0837	CAIS	Substitut	1 Nterm		143	427	p.(Lys143 *)							Sertoli-Leydig tumor	Female	Normal		Jazabeck et al. Gynecol Endocrinol 23: 499-504, 2007
0694	Prostate Cancer	Substitut	1 Nterm	*	144	431	p.(Gly144 Val)	16		normal	normal			Increased response to DHT	Male	Normal		Chen et al. The Prostate 63: 395-406, 2005
0516	CAIS	Substitut	1 Nterm		155	460	p.(Glu155 *)								Female	Normal		Copelli et al. Asian J Androl 1: 73-77, 1999
0523	CAIS	Substitut	1 Nterm		155	460	p.(Glu155 *)								Female	Normal		Gacobini et al. Hum Genet. 108. 176, 2001
0788	PAIS	Substitut	1 Nterm		159	475	p.(Ala159 Thr)											Mueller et al. Hum Genet 119: 681, 2006
0627	Prostate Cancer	Substitut	1 Nterm		168	502	p.(Gly168 Ser)							Orch + bicalutamide treatment Gleason 10 Somatic mutation	Male	Normal		Haapala et al. Lab Invest. 81: 1647-1651, 2001
0838	CAIS	Deletion	1 Nterm		168	502	p.(Gly168 fs)							1 nt deletion causing frameshift & stop	Female	Normal		Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0013	CAIS	Substitut	1 Nterm		174	521	p.(Leu174 *)								Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0316	PAIS	Substitut	1 Nterm		174	521	p.(Leu174 *)		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		174	521	p.(Leu174 *)	26	24	zero					Female	Normal	neg	Gottlieb et al. Hum Mutat. 14: 527-539, 1999
0566	Prostate cancer	Substitut	1 Nterm		178	534	p.(Ser178 )							Som mut. Estrogen treated. +p.Leu745 3350C>T silent mut	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0014	Prostate cancer	Substitut	1 Nterm		182	545	p.(Lys182 Arg)							Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0912	Prostate cancer	Substitut	1 Nterm		194	580	p.(Leu194 Phe)							Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442
0913	Prostate cancer	Substitut	1 Nterm		194	581	p.(Leu194 Arg)							Treated with anti-androgens	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442
0319	CAIS	Substitut	1 Nterm		196	587	p.(Gln196 Arg)							Also 1 nt deletion in Codon 598 causing a stop in codon 623	Female	Normal		Komori et al. J Obstetrics & Gynecol. 23: 277-81, 1997
0551	Prostate cancer	Substitut	1 Nterm		200	599	p.(Glu200 Gly)							Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0015	CAIS	Duplicat	1 Nterm		202	606	p.(Glu204 fs)			zero				4 nt insertion causing frameshift	Female	Normal	neg	Batch et al. Hum Mol Genet 1: 497, 1992
0549	Prostate cancer	Substitut	1 Nterm		204	612	p.(Glu204 Glu)							Treated with bicalutamide - silent mutation- somat. mut.	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0395	Normal	Substitut	1 Nterm		207	621	p.(Ser207 Arg)							Homosexual individual	Male	Normal		Macke et al. Am J Human Genetics 53: 844-852, 1993
0437	CAIS	Deletion	1 Nterm		210	619	p.(Arg210 fs)			zero				Frameshift	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0376	MAIS	Substitut	1 Nterm		212	636	p.(Arg212 Arg)							Silent mutation	Male	Normal		Wang et al. Clinical Genetics 54: 185-192, 1998
0328	Normal	Substitut	1 Nterm		213	639	p.(Glu213 )							Silent mutation - polymorphism detected in 8% popul.	Male	Normal		Batch et al. Hum Mol Genet 1: 497, 1992
0329	Normal	Substitut	1 Nterm		213	639	p.(Glu213 )							Silent mut.polymorph -detected in 14% of X chromosomes	Male	Normal		Hiort et al. Eur J Pediatr 153: 317-321, 1994
0330	Normal	Substitut	1 Nterm		213	639	p.(Glu213 )							Silent mutation polymorphism	Male	Normal		Lu et al. Clinical Genetics 49: 323-324. 1996



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								Amino acid	HGVS nt	Amino acid	Poly Gln #	Poly Gly #						Bmax
						Base	Base											
0917	Prostate cancer	Substitut	1 Nterm			229 1800	685 c.685A	p.(Thr229 Ala)						Both treated and untreated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0915	Prostate cancer	Indel	1 Nterm			229 1800	685 c.685_686AC	p.(Thr229 Cys)						Untreated - Occurred in two cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0916	Prostate cancer	Indel	1 Nterm			229 1800	685 c.685_686AC	p.(Thr229 Cys)						Both treated and untreated - Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0885	Prostate cancer	Indel	1 Nterm			229 1800	685 c.685_686AC	p.(Thr229 Cys)						+Gln58Leu, Ala253Val, Trp435Leu, Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0706	PAIS	Indel	1 Nterm	*		233 1812	697 c.697_698TC	p.(Ser233 Leu)			high	*			Male	Normal		Deeb et al. Clinical Endocrinol 63: 56-62, 2005
0531	MAIS	Substitut	1 Nterm	*		235 1820	705 c.715C	p.(Asn235 Lys)			normal	*		Azoospermia - transactivation 46% of wt	Male	Normal		Giwerzman et al. Clin Endocrinol 54: 827-834, 2001
0967	CAIS	Insertion	1 Nterm			241 1835	720 c.720_721	p.(Lys241 *)	21	18					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1030	MAIS	Substitut	1 Nterm			242 1838	724 c.721A	p.(Ala242 Ser)						Azoospermia	Male	Normal		Goglia et al. Fert Steril 96: 1165-1169, 2011
1049	PAIS	Substitut	1 Nterm			244 1844	730 c.730T	p.Ser244 Pro)						External Masculinisation Scale = 10	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012
0789	PAIS?	Substitut	1 Nterm			248 1858	743 c.743G	p.(Gly248 Val)							Male	Normal		Mueller et al. Hum Genet 119: 681, 2006
0918	Prostate Cancer	Substitut	1 Nterm			253 1873	758 c.758C	p.(Ala253 Val)						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			255 1878	763 c.763G	p.(Glu255 Lys)						+L446S, R485C, K610E, R787*, L798P, L874P, AR23, Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0350	CAIS	Substitut	1 Nterm	*		263 1903	788 c.788T	p.(Leu263 Pro)				*		Also Gly821Ala mut. Extra mutation causes greater thermolability	Female	Normal		Tanaka et al. Gynecol Endocrinol 12: 75-82, 1998
0845	CAIS?	Duplicat	1 Nterm			265 1908	793 c.793	p.(Gly265 fs)							Male	Normal		Mueller. Hum Genet 123: 105, 2008
0019	CAIS	Deletion	1 Nterm			266 1912	797 c.797	p.(Asp266 fs)			zero			1 nt deletion causing frameshift & stop in codon 296	Female	Normal		Bruggenwirth et al. J Steroid Biochem Mol Biol 58: 569-575, 1996
0017	Prostate cancer	Substitut	1 Nterm			268 1918	803 c.803T	p.(Met268 Thr)						Also p.Leu575Pro mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0018	Prostate cancer	Substitut	1 Nterm			271 1926	811 c.811C	p.(Pro271 Ser)						Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0780	MAIS	Substitut	1 Nterm			272 1929	814 c.814C	p.(Leu272 Phe)						Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0606	CAIS	Substitut	1 Nterm			289 1980	865 c.865G	p.(Glu289 *)			low	normal		low expression of WT AR - Somatic mosaicism	Female	Normal		Holterhaus et al. Genome Biology 4: R37
0556	Prostate cancer	Substitut	1 Nterm			298 2009	894 c.894C	p.(Ser298 Arg)						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			298 2009	894 c.894C	p.(Ser298 Arg)			normal			Transcrip activity activarted by estradiol and progesterone	Male	Normal		Li et al. Clin Exp Pharmacolo Physiol 35: 1252-1257, 2008
0816	Festicular cancer	Substitut	1 Nterm			299 2010	895 c.895G	p.(Ala299 Thr)						Seminoma	Male	Normal		Garolla et al. Encndorine Related Cancer 12: 645-655, 2005
1052	CAIS	Indel	1 Nterm			312 2048	933 c.933_1219 delins 77	p.(Phe312 fs)						287 deletion + 77 insertion causing frameshit and stop	Female	Normal	pos	Cong et al. Gene 500: 220-223, 2012
0550	Prostate cancer	Substitut	1 Nterm			336 2121	2121 c.1006T	p.(Ser336 Pro)						Treated with flutamide somatic mutation	Male	Normal		Taplin et al. 37th meeting ASCO 20: Abstr, 1738 2001
0398	Prostate cancer	Substitut	1 Nterm			342 2140	1025 c.1025C	p.(Pro342 Leu)						Somatic mutation Stage 3 tumor	Male	Normal		Castagnaro et al. Verh. Dtsch. Ges. Path. 77: 119-123, 1993







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									Bmax	Kd	Thermolabile k					
0782	MAIS	Substitut	1 Nterm	*	475	1424	p.(Ala475 Val)					male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0821	MAIS	Substitut	1 Nterm	*	475	1424	p.(Ala475 Val)	14	17			male infertility, low T severe hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
0822	MAIS	Substitut	1 Nterm	*	475	1424	p.(Ala475 Val)	24	17			male infertility, normal T	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
0823	MAIS	Substitut	1 Nterm	*	475	1424	p.(Ala475 Val)	19	17			male infertility, normal T, left cryptorchidism	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
0024	CAIS	Substitut	1 Nterm		481	1443	p.(Tyr481 *)	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		481	1443	p.(Tyr481 *)					Woolfian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0804	Liver cancer	Substitut	1 Nterm		481	1443	p.(Tyr481 )					Somatic mutation. Silent mutation	Female	Normal		Yeh et al. Int J Cancer 120: 1610-1617, 2007
0929	Prostate cancer	Substitut	1 Nterm		485		p.(Arg485 Cys)					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		485		p.(Arg485 Thr)					Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0546	CAIS	Deletion	1 Nterm		488		p.(Gln488 *)					3 affected siblings Variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0439	CAIS	Deletion	1 Nterm		489		p.(Gly489 fs)		low			5 nt deletion causing frameshift	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0440	CAIS	Substitut	1 Nterm		492	1474	p.(Gly492 Ser)		low	low			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0891	Prostate Cancer	Substitut	1 Nterm		498	1493	p.(Thr498 Ile)					Also p.Arg485Cys Bicalutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0931	Prostate Cancer	Substitut	1 Nterm		498	1493	p.(Thr498 Ile)					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		499	1495	p.(Ala499 Thr)					Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0933	Prostate Cancer	Substitut	1 Nterm		499	1496	p.(Ala499 Val)					Flutamide treated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0936	Prostate Cancer	Substitut	1 Nterm		500	1500	p.(Pro500 )					Treated with anti-androgens. Occurred in more than 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0025	CAIS	Substitut	1 Nterm		503	1508	p.(Trp503 *)						Female	Normal	pos	Bruggenwirth et al. J Steroid Biochem Mol Biol 58: 569-575, 1996
1071	CAIS	Substitut	1 Nterm		503	1508	p.(Trp503 *)						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0905	CAIS	Indel	1 Nterm		503	1507	p.(Trp503 fs)					Indel between c.1507_1512, 22 nt insertion	Female	Normal	neg	Philibert et al. Fertility & Sterility 94: 472-476 2010
0783	MAIS	Substitut	1 Nterm		507	1510	p.(Gly507 Asp)					Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0886	Prostate Cancer	Substitut	1 Nterm		509	1525	p.(Val509 Leu)					Bicalutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0934	Prostate Cancer	Substitut	1 Nterm		509	1525	p.(Val509 Leu)					Flutamide treated	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0935	Prostate Cancer	Substitut	1 Nterm		509	1526	p.(Val509 Gly)					Treated with anti-androgens. Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0339	MAIS	Substitut	1 Nterm	*	512	1536	p.(Val512 Val)					Oligospermia caused by silent mutation	Male	Normal		Hiort et al. 80th US Endo Soc Meeting. Abstr P2-38, 1998









Accession #	Phenotype	Mutation type	Exon Domain	CpG spot	Pathogenicity proven	Amino acid Base	Position HGVS nt	Change Amino acid Base	Exon 1 tracts	Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
										Bmax	Kd	Thermolabile k					
0610	Normal	Substitut	3 DBD	598		1794	p.(Ser598 Arg)	2909 c.1794C					Adult had bilateral cryptorchadism in childhood	Male	Normal		Giweremann et al. Horm Res 61: 58-62, 2004
0959	E006AA Prost Can cell line	Substitut	3 DBD	598		1792	p.(Ser598 Gly)	2907 c.1792A					Dominant-negative loss of function mut AR gene amplificat	Male	Normal		D'Antonio et al. PLOS One 5: e11475, 2010
0720	CAIS	Deletion	3 DBD	600		1799	p.(Asn600 fs)	2914 c.1799A					1 nt deletion causing frameshift & stop at codon 625	Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0055	CAIS	Substitut	3 DBD	602		1805	p.(Cys602 Phe)	2920 c.1805G						Female	Normal	pos	Baldazzi et al. Hum Mol Genet 3: 1169-70 1994
0946	PAIS	Substitut	3 DBD	602	*	1805	p.(Cys602 Ser)	2920 c.1805G					40% loss of ligand-binding & 70% loss of transactivation	Female	Ambiguous		Singh et al. J Steroid Biochem Mol Biol 2010
1075	CAIS	Substitut	3 DBD	602	*	1805	p.(Cys602 Tyr)	2920 c.1805G						Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0362	PAIS	Substitut	3 DBD	603		1807	p.(Thr603 Pro)	2922 c.1807A	24				testis located in inguinal canal. Same family as 0831	Male	Ambiguous	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0831	PAIS	Substitut	3 DBD	603		1807	p.(Thr603 Pro)	2922 c.1807A					Same family as 0362	Male	Ambiguous	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0942	PAIS	Substitut	3 DBD	604		1811	p.(Ile604 Asn)	2926 c.1811T		zero			Somatic mosaicism - no transcriptional activity				Elfferich et al. Sexual Development 3: 237-244, 2009
1034	CAIS	Substitut	3 DBD	604		1811	p.(Ile604 Ser)	2926 c.1811T					Twins	Male	Normal		Hughes et al., The Lancet 380:1419-1428, 2012
0056	PAIS	Substitut	3 DBD	605		1813	p.(Asp605 Tyr)	2928 c.1813G						Male	Ambiguous		Hiort et al: Hum Mol Genet 3: 1163-1166 1994
0598	PAIS	Substitut	3 DBD	605		1813	p.(Asp605 Tyr)	2928 c.1813G						Female	Ambiguous	pos	Scheiber et al. J Ped Endocrinol & Metab. 16: 367-373, 2003
1074	PAIS	Substitut	3 DBD	605		1813	p.(Asp605 Asn)	2928 c.1813G									Hughes et al. Semin Reprod Med 30: 432-442, 2012
0848	CAIS	Substitut	3 DBD	608	*	1822	p.(Arg608 *)	2937 c.1822C					Diag at 1 Mo-Wolffian derivatives on rt side only	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0057	CAIS	Substitut	3 DBD	608	*	1822	p.(Arg608 *)	2937 c.1822C		zero				Female	Normal		Brown et al. Eur J Pediatr (Suppl 2) 152. S62, 1993
0511	CAIS	Substitut	3 DBD	608	*	1822	p.(Arg608 *)	2937 c.1822C		zero				Female	Normal		Giwereman et al. Hormone Research 53: 83-88, 2000
0702	CAIS	Substitut	3 DBD	608	*	1822	p.(Arg608 *)	2937 c.1822C					1 affected sibling	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0770	MAIS	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G					Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0677	PAIS	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G							Ambiguous		Bouvattier et al: J Clin Endocrinol & Metab 87: 29-32, 2002
0393	PAIS	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G					Germ cell tumour - in undescended testis	Female	Normal		Chen et al. Human Reproduction 14: 664-670, 1999
0347	PAIS	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G					Patient successfully treated with testosterone	Male	Ambiguous		Weidemann et al. J Clin Endocrinol & Metab 83: 1173-1176, 1998
0060	PAIS	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G						Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0059	PAIS	Substitut	3 DBD	608	**	1823	p.(Arg608 Gln)	2938 c.1823G		normal normal				Male	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733-739, 1996
0058	PAIS and breast cancer	Substitut	3 DBD	608	*	1823	p.(Arg608 Gln)	2938 c.1823G						Male	Ambiguous	pos	Wooster et al. Nat Genet 2: 132, 1992
0847	CAIS	Substitut	3 DBD	608		1822	p.(Arg608 *)	2937 c.1822C					Diag at 1Mo Bilateral gonadectomy at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008







Accession #	Phenotype	Mutation type	Exon Domain	Pathogenicity proven CpG hot spot	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
								Poly Gln #	Poly Gly #	Bmax	Kd	k						
0812	PAIS	Substitut	4 LBD	*	646	1937	p.(Ala646 Asp)	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	*	646	1937	p.(Ala646 Asp)	30						Long poly Q and short poly G contribute to PAIS	Male	Ambiguous		Werner et al. J Clin Endocrinol Metab 91: 3515-3520, 2006
1078	PAIS	Substitut	4 LBD		646	1937	p.(Ala646 Asp)	30										Hughes et al. Semin Reprod Med 30: 432-442, 2012
0081	Prostate cancer	Substitut	4 LBD		648	1943	p.(Ser648 Asn)							Also p.G725D, L881Q & A897T mut Somatic mutations	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
1020	Premature ovarian failure	Substitut	4 LBD		650	1948	p.(Thr650 Ala)							Patient had menopause at 28	Female	Normal		Panda et al. Gynecol Endocrinol 27:1-7, 2011
0576	Prostate cancer	Substitut	4 LBD		650	1950	p.(Thr650 Thr)							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		651	1951	p.(Ser651 Gly)							Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0827	MAIS	Substitut	4 LBD	*	651	1951	p.(Ser651 Gly)	23	18					Hypoandrogenism, scrotal hypoplasia hypospematogenesis	Male	Normal		Zuccarello et al. Clin Endocrinol 68: 58-588, 2008
0555	PAIS	Substitut	4 LBD		654	1960	p.(Glu654 Lys)	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		658	1972	p.(Gln658 *)								Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001
1021	Premature ovarian failure	Substitut	4 LBD		658	1972	p.(Gln658 Lys)							Patient had menopause at 17	Female	Normal		Panda et al. Gynecol Endocrinol 27: 1-7, 2011
0082	PAIS	Substitut	4 LBD		665	1994	p.(Ile665 Asn)	22	22	low		norm						Pinsky et al. Clin Inv Med 15: 456, 1992
1079	PAIS	Substitut	4 LBD		665	1994	p.(Ile665 Asn)											Hughes et al. Semin Reprod Med 30: 432-442, 2012
0937	Prostate cancer	Substitut	4 LBD		666		p.(Glu666 Asp)							Treated with anti-androgens Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
1083	CAIS	Substitut	4 LBD		671	2011	p.(Gln *)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0083	Prostate cancer	Substitut	4 LBD		671	2012	p.(Gln671 Arg)							Also p.S792P 3489T>C mut. Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
0084	PAIS	Substitut	4 LBD		672	2015	p.(Pro672 His)								Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0943	CAIS	Substitut	4 LBD	*	672	2014	p.(Pro672 Ser)							Reduced transactivat no change in DNA or FxxLF binding	Female	Normal		Elfferich et al. Sexual Development 3: 237-244, 2009
0085	Prostate cancer	Substitut	4 LBD		673	2018	p.(Ile673 Thr)							Somatic mutation	Male	Normal		Tilley et al. Clinical Cancer Res. 2: 277-285, 1996
1079	PAIS	Substitut	4 LBD		674	2021	p.(Phe674 Cys)											Hughes et al. Semin Reprod Med 30: 432-442, 2012
0086	CAIS	Substitut	4 LBD		678	2033	p.(Leu678 Pro)			zero					Female	Normal	pos	Belsham et al. Human Mutation 5: 28-33, 1995
1080	PAIS	Substitut	4 LBD		679	2036	p.(Glu679 Gly)											Hughes et al. Semin Reprod Med 30: 432-442, 2012
0723	CAIS	Substitut	4 LBD		681	2042	p.(Ile681 Thr)							Affected twin sister	Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005
1005	PAIS	Substitut	4 LBD		681	2042	p.(Ile681 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
0087	CAIS	Substitut	4 LBD		682	2044	p.(Glu682 Lys)								Female	Normal		Hiort et al. J Clin Endocrinol Metab 77: 262-266, 1993

Accession #	Phenotype	Mutation type	Exon Domain	Pathogenicity proven CpG spot	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference	
								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k							
0394	CAIS	Substitut	4 LBD		682	2044	p.(Glu682 Lys)							Germ cell tumour in undescended testis	Female	Normal		Chen et al. Human Reproduction 14: 664-670, 1999	
0618	CAIS	Substitut	4 LBD		682	2044	p.(Glu682 *)	21			v low				Female	Normal		MacLean et al. Hum Mutat 23: 287, 2004	
0619	CAIS	Substitut	4 LBD		682	2044	p.(Glu682 *)	21			low				Female	Normal		MacLean et al. Hum Mutat 23: 287, 2004	
947	CAIS	Substitut	4 LBD		682	2046	p.(Glu682 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	
0534	PAIS	Substitut	4 LBD		683	2047	p.(Pro683 Thr)				low				Female	Ambiguous		Chavez et al. J Hum Genet. 46: 560-565, 2001	
0724	CAIS	Substitut	4 LBD		683	2047	p.(Pro683 Ala)				low			aunt, 2 sisters and neice affected	Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005	
0089	Prostate cancer	Substitut	4 LBD		684	2051	p.(Gly684 Ala)							Somatic mutation - transactivation normal	Male	Normal		Koivisto et al. Cancer Research 57: 314-319, 1997	
0090	CAIS	Substitut	4 LBD		685	2053	p.(Val685 Ile)				zero				Female	Normal		Mebarki et al. 75th US Endo Soc Meeting, Abstr 602, 1993	
0945	CAIS	Duplicat	4 LBD		686	2055	p.(Val686 fs)							2 nt duplicat. causing frameshift & stop in Codon 788	Female	Normal	pos	Rong et al. Eur J Obst & Gynecol & Reprod Biol 148: 53-55, 2010	
0091	PAIS	Substitut	4 LBD		687	2059	p.(Cys687 Arg)								Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996	
1081	PAIS	Substitut	4 LBD		688	2063	p.(Ala688 Asp)									Ambiguous		Hughes et al. Semin Reprod Med 30: 432-442, 2012	
0092	PAIS	Substitut	4 LBD		688	2063	p.(Ala688 Val)								Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996	
0093	CAIS	Substitut	4 LBD		689	2066	p.(Gly689 Glu)							de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998	
0446	CAIS	Substitut	4 LBD		689	2065	p.(Gly689 *)				zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000	
0575	CAIS	Substitut	4 LBD	*	690	2069	p.(His690 Pro)				v low	low	high	* v. low transactivation activity	Female	Normal		Rosa et al. J Clin Endocrinol Metab 87: 4378-4382, 2002.	
0094	PAIS	Deletion	4 LBD		691	2071	p.(Asp691 del)									Normal		pos	Schwartz et al. Horm Res 41: 117 Abstr 244, 1994
0656	CAIS	Substitut	4 LBD	*	691	2072	p.(Asp691 Val)					high		Epididymis & Vas deferens present - v low transactivation	Female	Normal		Tadokoro et al. Clinical Endocrinology 71: 253-260, 2009	
1082	PAIS	Substitut	4 LBD		691	2073	p.(Asp691 Glu)									Ambiguous		Hughes et al. Semin Reprod Med 30: 432-442, 2012	
0095	CAIS	Deletion	4 LBD		693	2077	p.(Asn693 del)				normal	high		* Three nucleotide deletion	Female	Normal		Batch et al. Hum Mol Genet 1: 497-503, 1992	
0704	PAIS	Deletion	4 LBD		693	2077	p.(Asn693 del)				normal	high		3 nt deletion. 3 affected siblings . variable phenotype	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001	
0849	CAIS	Substitut	4 LBD		694	2080	p.(Gln694 *)							Diag at 17y- Bilateral gonadectomy 17y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008	
0096	CAIS	Substitut	4 LBD	*	696	2086	p.(Asp696 His)				low				Female	Normal	neg	Ris-Stalpers et al. Mol Endocrinol 5: 1562, 1991	
0097	CAIS	Substitut	4 LBD	**	696	2086	p.(Asp696 Asn)				normal	normal	high	mutation found in two unrelated families	Female	Normal	pos	Ris-Stalpers et al. Mol Endocrinol 5: 1562, 1991	
0098	PAIS	Substitut	4 LBD	*	696	2086	p.(Asp696 Asn)							de novo mutation	Female	Ambiguous		Hiort et al. J Pediatrics 132: 939- 943, 1998	
0657	CAIS	Substitut	4 LBD	*	696	2086	p.(Asp696 Asn)							Wolffian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004	

Accession #	Phenotype	Mutation type	Exon Domain	CpG spot	Pathogenicity proven	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
									Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0773	MAIS	Substitut	4 LBD			696	2086	p.(Asp696 Asn)								Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0977	CAIS	Substitut	4 LBD			696	2086	p.(Asp696 Asn)								Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0978	CAIS	Substitut	4 LBD			696	2086	p.(Asp696 Asn)	22	17						Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0977	CAIS	Substitut	4 LBD			696	2086	p.(Asp696 Tyr)	19	18						Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0335	CAIS	Substitut	4 LBD			696	2087	p.(Asp696 Val)		21						Female	Normal	pos	Dork et al. Human Mutation 11: 337-339, 1998
1053	CAIS	Substitut	4 LBD			698	2094	p.(Phe698 Leu)								Female	Normal		Ning et al. Urology 80: 216-218, 2012
0447	CAIS	Substitut	4 LBD			701	2101	p.(Leu701 Met)			zero					Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0725	CAIS	Deletion	4 LBD			701	2101	p.(Leu701 fs)								Female	Normal	pos	Ledig et al. Horm Res 63: 263-269, 2005
1084	CAIS	Substitut	4 LBD			701	2103	p.(Leu701 Phe)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0448	CAIS	Substitut	4 LBD			702	2094	p.(Leu702 Phe)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0518	PAIS	Substitut	4 LBD			702	2104	p.(Leu702 Ile)											Chavez et al. Clin Genet 59: : 185-188, 2001
0099	Prostate cancer	Substitut	4 LBD			702	2105	p.(Leu702 His)								Male	Normal		Suzuki et al. J Steroid Biochem Molec Biol 46: 759, 1993
0326	Prostate cancer	Substitut	4 LBD			702	2105	p.(Leu702 His)								Male	Normal		Watanabe et al. Jpn J Clin Oncol 27: 389-393, 1997
0408	MDA PCa-Za	Substitut	4 LBD			702	2105	p.(Leu702 His)			normal low					Male	Normal		Zao et al. J of Urology 162: 2192-2199, 1999
0100	CAIS	Substitut	4 LBD			703	2107	p.(Ser703 Ala)			zero					Female	Normal		Pinsky et al. Clin Inv Med 15: 456-472, 1992
0101	PAIS	Substitut	4 LBD			704	2110	p.(Ser704 Gly)			low	high				Male	Ambiguous		Radnays et al. J of Urology 158: 1553-1556, 1997
0449	CAIS	Substitut	4 LBD			704	2110	p.(Ser704 Gly)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0712	PAIS	Substitut	4 LBD			704	2110	p.(Ser704 Gly)				high				Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56-62, 2005
0713	PAIS	Substitut	4 LBD			704	2110	p.(Ser704 Gly)								Female	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56-62, 2005
0979	CAIS	Substitut	4 LBD			704	2110	p.(Ser704 Cys)	19	18	low	normal				Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1085	CAIS	Substitut	4 LBD			704	2110	p.(Ser704 Cys)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
1108	CAIS	Substitut	4 LBD			704	2111	p.(Ser704 Ile)								Female	Normal		Chin et al. J Pediatr Endocrinol Metab 25:1145-1051, 2012
0102	CAIS	Substitut	4 LBD			706	2117	p.(Asn706 Ser)			zero					Female	Normal		Pinsky et al. Clin Inv Med 15: 456-472, 1992
0103	CAIS	Substitut	4 LBD			706	2117	p.(Asn706 Ser)			zero					Female	Normal		DeBellis et al. Mol Endocrinol 6: 1909-20, 1992
0104	CAIS	Substitut	4 LBD			706	2117	p.(Asn706 Ser)								Female	Normal		Quigley et al. Endocrine Reviews 16: 271, 1995



Accession #	Phenotype	Mutation type	Exon Domain	Pathogenicity proven CpG spot	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
										Bmax	Kd	Thermolabile k					
0505	PAIS	Substitut	4 LBD	*	713	2137	p.(Leu713 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
0506	PAIS	Substitut	4 LBD	*	713	2137	p.(Leu713 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
0507	PAIS	Substitut	4 LBD	*	713	2137	p.(Leu713 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
0109	Prostate cancer	Substitut	4 LBD	**	716	2146	p.(Val716 Met)			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	*	716	2146	p.(Val716 Met)			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	*	716		p.(Val716 *)						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endocrinol & Metab 87: 29-32, 2002
0792	PAIS	Substitut	4 LBD		719	2155	p.(Trp719 Arg)						Somatic mosaic 2/3 mutant to 1/3 wt				Mueller et al. Hum Genet 119: 680, 2006
0111	CAIS	Substitut	4 LBD		719	2157	p.(Trp719 *)			zero				Female	Normal	pos	Sai et al. Am J Hum Genet 46: 1095, 1990
1088	CAIS	Substitut	4 LBD		719	2157	p.(Trp719 *)							Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0112	Prostate cancer	Substitut	4 LBD		721	2161	p.(Lys721 Glu)						Somatic mutation- Bone metastases of Prostate cancer	Male	Normal		Kleinerman et al. J of Urology 155: 624A, 1996
0113	Prostate cancer	Substitut	4 LBD		722	2164	p.(Ala722 Thr)						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		722	2164	p.(Ala722 fs)						1 nt frameshift & stop in codon 767 - no immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0584	CAIS	Deletion	4 LBD		722	2166	p.(Ala722 fs)						1 nt frameshift & stop - low immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0114	CAIS	Substitut	4 LBD		723	2169	p.(Leu723 Phe)							Female	Normal		Hiort et al: Am J Med Genet. 63: 218-222, 1996
0761	Prostate cancer	Substitut	4 LBD		723	2169	p.(Leu723 Phe)						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		724	2170	p.(Pro724 Ser)			normal	high		-Epididymis present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0791	PAIS ?	Substitut	4 LBD		724	2171	p.(Pro724 Leu)										Mueller et al. Hum Genet 119: 681, 2006
0452	CAIS	Substitut	4 LBD		725	2173	p.(Gly725 Ser)			zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0453	CAIS	Deletion/ Insertion	4 LBD		725	2174	p.(Gly725 Asp)			zero			Epididymis & Vas deferens present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0620	CAIS	Substitut	4 LBD		725	2174	p.(Gly725 Ala)	19		normal				Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
0798	CAIS	Substitut	4 LBD	*	725	2174	p.(Gly725 Val)			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
1107	CAIS	Deletion	4-8 LBD											Female	Normal		
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							zero			p.(Tyr>Arg) substitution	Female	Normal		Marcelli et al. 74th US Endo Soc Meetings: Abstr. 224, 1992

Accession #	Phenotype	Mutation type	Exon Domain	CpG spot	Pathogenicity proven	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
									Poly Gln #	Poly Gly #	Bmax	Kd	k	Thermolabile					
0118	PAIS	Substitut	5 LBD	*		726 3291	2176 c.2176T	p.(Phe726 Leu) 3291T>C				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			726 3291	2176 c.2176T	p.(Phe726 Leu) 3291T>C							Hypospadias and cryptorchidism	Male	Ambiguous	pos	Nordenskjold et al Urological Res, 27: 49 -55, 1999
0119	Prostate cancer	Substitut	5 LBD	*		727 3295	2180 c.2180G	p.(Arg727 Leu) 3295G>T				normal	normal		Germ line mutation present in offspring	Male	Normal	pos	Elo et al. J Clin Endocrinol Metab, 80: 3494-3500, 1995
0508	Prostate cancer	Substitut	5 LBD	*		727 3295	2180 c.2180G	p.(Arg727 Leu) 3295G>T							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			727 3295	2180 c.2180G	p.(Arg727 Leu) 3295G>T	20						Somatic mut polyGln 24 to 20 Orchiectomy Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0687	Normal	Substitut	5 LBD			727 3295	2180 c.2180G	p.(Arg727 Leu) 3295G>T							Patient suffering from alcoholism		Normal		Yan et al. Psychiatric Genet 14: 57-60, 2004
0120	MAIS	Substitut	5 LBD			728 3299	2184 c.2184C	p.(Asn728 Lys) 3299C>G							Oligospermia	Male	Normal		Yong et al. Lancet, 344: 826-827, 1994
0121	PAIS	Substitut	5 LBD			729 3301	2186 c.2186T	p.(Leu729 Ser) 3301T>C		low			*						McPhaul et al. J Clin Inv, 90: 2097, 1992
0122	Prostate Cancer	Substitut	5 LBD	*		731 3306	2191 c.2191G	p.(Val731 Met) 3306G>A							Somatic mutation	Male	Normal		Newmark et al. Proc Natl AcadSci 89: 6319, 1992
0123	Prostate Cancer	Substitut	5 LBD	*		731 3306	2191 c.2191G	p.(Val731 Met) 3306G>A							Somatic mutation	Male	Normal		Petersiel et al. Int J Cancer 63: 544-550, 1995
0762	Prostate Cancer	Substitut	5 LBD	*		731 3306	2191 c.2191G	p.(Val731 Met) 3306G>A							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	*		733 3312	2197 c.2197G	p.(Asp733 Tyr) 3312G>T			high						Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0126	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Tyr) 3312G>T		zero							Normal		Pinsky et al. Clin Inv Med 15: 456, 1992
0127	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Tyr) 3312G>T									Normal		Ghirri and Brown. Pediatr Res 33: Abstr 95, 1993
0801	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Tyr) 3312G>T							6 affected family members		Normal	pos	Scott et al. Endocr Pract 12: 664-669, 2006
0124	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Asn) 3312G>A			high						Normal		Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0659	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Asn) 3312G>A							Epididymis & Vas deferens present		Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0310	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Asn) 3312G>A	19								Normal		Ko et al. J Reprod. Med 42: 424- 427, 1997
0955	CAIS	Substitut	5 LBD			733 3312	2197 c.2197G	p.(Asp733 Asn) 3312G>A									Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
0628	Prostate cancer	Substitut	5 LBD			733 3314	2199 c.2199C	p.(Asp733 Asp) 3314C>T							Orch + Bicalutamide +phosphamide treat Gleason 10. Somatic	Male	Normal		Haapala et al. Lab Invest. 81: 1647-1651, 2001
0752	CAIS	Substitut	5 LBD			734 3315	2200 c.2200C	p.(Gln734 *) 3315C>T							No WD development		Normal	neg	Barbaro et al. Clin Endocrinol 66: 822 -826, 2007
0128	PAIS	Substitut	5 LBD			734 3317	2202 c.2202G	p.(Gln734 His) 3317G>T							This patient was a mosaic for wt. & mut. alleles- de novo mut.		Ambiguous	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
0726	CAIS	Substitut	5 LBD			738 3327	2212 c.2212A	p.(Ile738 Phe) 3327A>T									Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0129	PAIS	Substitut	5 LBD	*		738 3328	2213 c.2213T	p.(Ile738 Thr) 3328T>C		low					Mutation disrupts N/C terminal interaction	Male	Ambiguous	pos	Quigley et al. Mech Aging & Develop 125: 683-695
0944	CAIS	Substitut	5 LBD	*		739 3331	2216 c.2216A	p.(Gln739 Arg) 3331A>G							V reduced transactiv reduced interaction with FxxLX		Normal		Elfferich et al. Sexual Development 3: 237 -244, 2009

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0530	CAIS	Substitut	5 LBD	*	740	2218	p.(Tyr740 Asp)				zero			no transactivation in COS-1 cells	Female	Normal		Suzuki et al. Int. J Andrology 24: 183-188, 2001
0616	PAIS	Substitut	5 LBD	*	741	2222	p.(Ser741 Cys)				v low			Acute stress masked PAIS.	Male	Ambiguous	pos	Pitteloud et al. J Clin Endocrinol & Metab 89: 1053-1058, 2004
0130	CAIS	Substitut	5 LBD	*	742	2224	p.(Trp742 Arg)				low				Female	Normal	neg	Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0360	Prostate cancer	Substitut	5 LBD		742	2225	p.(Trp742 *)							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0595	CAIS	Substitut	5 LBD	*	742	2225	p.(Trp742 Leu)							Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0858	CAIS	Substitut	5 LBD		742	2225	p.(Trp742 Leu)							Diag at birth bilateral gonadectomy at 14yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0859	CAIS	Substitut	5 LBD		742	2225	p.(Trp742 Leu)							Diag at birth bilateral gonadectomy at 12.5yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0629	Prostate cancer	Substitut	5 LBD		742	2226	p.(Trp742 Cys)							Orch + bicalutamide Gleason 10- somatic mutation	Male	Normal		Haaplaa et al. Lab Invest. 81: 1647-1651, 2001
0635	PAIS	Substitut	5 LBD		742	2226	p.(Trp742 Cys)		27	low	high			Testes located in scrotum - Also has p. (Leu548Phe mutation	Male	Ambiguous	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0552	Prostate cancer	Substitut	5 LBD		742	2226	p.(Trp742 Cys)							Treated with bicalutamide - somatic mutation	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673-2678, 2003
0982	CAIS	Substitut	5 LBD		742	2224	p.(Trp742 Arg)		23	17					Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0131	PAIS	Substitut	5 LBD		743	2227	p.(Met743 Val)					high						Ris-Stalpers et al. Pediatric Res. 36: 227-234,1994
0341	PAIS	Substitut	5 LBD		743	2227	p.(Met743 Val)		21	zero				Testis located in inguinal region	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0744	PAIS	Substitut	5 LBD		743	2227	p.(Met743 Val)							affected niece of 0131	Female	Ambiguous		Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0132	PAIS	Substitut	5 LBD	*	743	2229	p.(Met743 Ile)				normal	high	*		Female	Ambiguous		Batch et al. Hum Mol Genet 1: 497, 1992
0948	PAIS	Substitut	5 LBD		743	2229	p.(Met743 Ile)								Female	Ambiguous		Nagaraja et al. J Pediatr Endocrinol Metab 22: 1169-1173, 2009
1006	PAIS	Substitut	5 LBD		743	2229	p.(Met743 Ile)		18	18	low	normal		Mother heterozygote carrier	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0133	PAIS	Substitut	5 LBD	*	744	2229	p.(Gly744 Val)				low	high		Transcription only at high conc of androgen	Female	Ambiguous		Georget et al. J Clin Endocrinol Metab 83: 3597-3603, 1998
0134	PAIS	Substitut	5 LBD		744	2229	p.(Gly744 Val)				normal	normal	*					Nakao et al. J Clin Endocrinol Metab 77: 103-107, 1993
0414	CAIS	Substitut	5 LBD		744	2229	p.(Gly744 Val)				zero			de novo mutation	Female	Normal		Lobaccaro et al. J Steroid Biochem & Mol Biol. 44: 211-216, 1993
0536	CAIS	Substitut	5 LBD		744	2229	p.(Gly744 Glu)				normal				Female	Normal		Chavez et al. J Hum Genet. 46: 560-56, 2001
0564	CAIS	Substitut	5 LBD	*	744	2229	p.(Gly744 Glu)				v low	high	high	*	Female	Normal		Pujol et al. J Clin Endocrinol & Metab. 87: 5793-5800, 2002
0572	Prostate cancer	Substitut	5 LBD		744	2232	p.(Gly744 )							Somatic mutation + estrogen treatment Horm-refractory CaP	Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
0361	Prostate cancer	Deletion	5 LBD		744	2232	p.(Gly744 fs)							Frameshift-somatic mut.- separate tumor in same indiv. as 0362	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0519	CAIS	Substitut	5 LBD		744	2230	p.(Gly744 Arg)								Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0983	CAIS	Substitut	5 LBD		744	2231	p.(Gly744 Val)	24	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0135	CAIS	Substitut	5 LBD		745	2233	p.(Leu745 Phe)						1 affected sibling				pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0984	CAIS	Substitut	5 LBD		745	2233	p.(Leu745 Phe)	21	17				1 heterozygote sister & 1 wt sister	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0362	Prostate cancer	Substitut	5 LBD		745	2233	p.(Leu745 Phe)						Somatic mutation - separate tumor in same indiv. as 0361	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995	
0136	PAIS	Substitut	5 LBD		746	2237	p.(Met746 Thr)			zero								Ris-Stalpers et al. Pediatric Res 36: 227-234, 1994
0727	CAIS	Substitut	5 LBD		746	2237	p.(Met746 Thr)						Mother and sisters heterozygous	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005	
0755	CAIS	Substitut	5 LBD	*	746	2238	p.(Met746 Ile)						Enhanced affinity for & increased transact for estradiol	Female	Normal		Bonagura et al. Mol Cell Endocrinol 263: 79-89, 2007	
1007	PAIS	Substitut	5 LBD		746	2236	p.(Met746 Leu)	23	17				mother & grandmother heterozygote carriers	Male	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0138	PAIS	Substitut	5 LBD		747	2239	p.(Val747 Met)							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996	
0137	PAIS	Substitut	5 LBD		747	2239	p.(Val747 Met)											Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0949	PAIS	Substitut	5 LBD		747	2239	p.(Val747 Met)											Nagaraja et al. J Pediatr Endocrinol Metab 22: 1169-1173, 2009
0785	MAIS	Substitut	5 LBD		748	2242	p.(Phe748 Ile)						Male Infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006	
0985	CAIS	Substitut	5 LBD		748	2243	p.(Phe748 Cys)	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		748	2244	p.(Phe748 fs)						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		749	2245	p.(Ala749 Thr)						Also p.(S866P; Q868* and Q920R). Somatic mutations	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000	
0565	Prostate cancer	Substitut	5 LBD	*	749	2245	p.(Ala749 Thr)			normal	high		Somatic mutation. weaker interaction with HSP	Male	Normal		James et al. Mol Endocrinol. 16: 2692-2705, 2002	
0139	PAIS	Substitut	5 LBD	*	749	2246	p.(Ala749 Asp)			low	high		Abnormal dissociation				Marcelli et al. J Clin Invest 94: 1642-1650, 1994	
0363	Prostate cancer	Substitut	5 LBD		749	2246	p.(Ala749 Val)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995	
0140	CAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)							Female	Normal	pos	DeBellis et al. Mol Endocrinol 6: 1909-20, 1992	
0141	CAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)							Female	Normal	pos	Jakubiczka et al. Hum Genet 90: 311-2, 1992	
0483	PAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)			normal high								Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0614	CAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)						Fallopian tube present	Female	Normal		Van et al. Eur J. Pediatr. 162: 781-784, 2003	
0679	CAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)						bilateral inguinal hernia	Female	Normal		Bouvattier et al: J Clin Endocrinol & Metab 87: 29-32, 2002	
0680	CAIS	Substitut	5 LBD		750	2248	p.(Met750 Val)						Diff between ext genital & chrom - in amniocentesis	Female	Normal		Bouvattier et al: J Clin Endocrinol & Metab 87: 29-32, 2002	
0364	Prostate cancer	Substitut	5 LBD		750	2250	p.(Met750 Ile)						Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995	



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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0630	Prostate cancer	Substitut	5 LBD		750	2250	p.(Met750 Ile)							Orch + bicalutamide Gleason 10 +silent mut Q868Q(G>A)	Male	Normal		Haaplaa et al. Lab Invest. 81: 1647-1651, 2001
0365	Prostate cancer	Substitut	5 LBD		751	2251	p.(Gly751 Ser)							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0142	CAIS	Substitut	5 LBD	*	751	2252	p.(Gly751 Asp)				v low			Mutation found in two unrelated patients	Female	Normal		Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
0143	CAIS	Substitut	5 LBD		751	2252	p.(Gly751 Asp)								Female	Normal		Brown et al: 74th US Endo Soc Meeting Abstr 1506, 1992
0144	CAIS	Substitut	5 LBD		752	2254	p.(Trp752 Arg)				low	high		Sibling of 0703	Female	Normal	pos	Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
0366	Prostate cancer	Substitut	5 LBD		752	2255	p.(Trp752 *)							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0367	Prostate cancer	Substitut	5 LBD		752	2255	p.(Trp752 *)							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0368	Prostate cancer	Substitut	5 LBD		752	2256	p.(Trp752 *)							Somatic mutation	Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
0401	CAIS	Substitut	5 LBD		752	2256	p.(Trp752 *)				zero				Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263-272, 1999
0664	PAIS	Substitut	5 LBD		752	2256	p.(Trp752 *)							Somatic mosaicism - male changed to female at 18mths	Female	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0703	CAIS	Substitut	5 LBD		752	2254	p.(Trp752 Arg)				normal	high		Sibling of 0144	Female	Normal		Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
1089	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0145	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)				zero				Female	Normal		Pinsky et al. Clin Inv Med 15: 456, 1992
0146	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)								Female	Normal		Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
0342	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)				16	zero		Testes located in abdomen	Female	Normal	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0402	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)					zero			Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263-272, 1999
0728	CAIS	Substitut	5 LBD	*	753	2257	p.(Arg753 *)								Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0754	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)							Inherited from maternal grandmother present in two sisters	Female	Normal	pos	Olafsson et al. Laeknabladid 86: 263-166, 2000
0147	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 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
0333	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)								Female	Normal	pos	Komori et al. Arch Gynecol & Obstetrics 261: 95-100, 1998
0349	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)								Female	Normal		Cabral et al. Brazilian J Med & Biol Res. 31: 775-758, 1998
0148	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)					zero		Equivalent to tfm rat	Female	Normal		Evans. J Endocrinol 135 Suppl, Abstr P26, 1992
0497	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)							Bilateral testicular tumors	Female	Normal		Sakai et al. IntJ of Urology 7: 390-392, 2000
0860	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)							Prenatal diagnosis	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
1090	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Gln)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
1091	CAIS	Substitut	5 LBD	*	753	2258	p.(Arg753 Pro)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
					3373	c.2258G	3373G >C											
0149	CAIS	Substitut	5 LBD		755	2263	p.(Phe755 Val)		zero						Female	Normal		Lobaccaro et al. Hum Mol Genet 2: 1041-1043, 1993
					3378	c.2263T	3378T >G											
0369	Prostate cancer	Substitut	5 LBD		755	2263	p.(Phe755 Leu)						Somatic mutation		Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
					3378	c.2263T	3378T >C											
0879	CAIS	Substitut	5 LBD	*	755	2263	p.(Phe755 Val)		high				Greatly reduced transactivation 5%		Female	Normal		Tadokoro et al. Clinical Endocrinology 71: 253-260, 2009
					3378	c.2263T	3378T >G											
0150	CAIS	Substitut	5 LBD		755	2263	p.(Phe755 Val)								Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
					3378	c.2263T	3378T >G											
0880	PAIS	Substitut	5 LBD	*	755	2264	p.(Phe755 Ser)		normal				Microphallus no hypospadias		Male	Ambiguous		Tadokoro et al. Clinical Endocrinology 71: 253-260, 2009
					3379	c.2264T	3379T >C											
1050	MAIS	Substitut	5 LBD	*	755	2264	p.(Phe755 Ser)						Azoospermia. Fertile - intracytoplasmic sperm injection		Male	Normal		Massin et al. Clin Endocrinol (Oxf) 77:593-598, 2012
					3379	c.2264T	3379T >C											
0715	PAIS	Substitut	5 LBD	*	755	2265	p.(Phe755 Leu)		high				Microphallus, hypospadias, cryptorchidisms		Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56-62, 2005
					3380	c.2265C	3380C >A											
0152	PAIS	Substitut	5 LBD	*	755	2265	p.(Phe755 Leu)		normal high			*			Male	Ambiguous		Weidemann et al. Clin Endocrinology 45: 733-739, 1996
					3380	c.2265C	3380C >A											
0151	PAIS	Substitut	5 LBD		755	2265	p.(Phe755 Leu)								Male	Ambiguous		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
					3380	c.2265C	3380C >A											
0714	PAIS	Substitut	5 LBD		755	2265	p.(Phe755 Leu)		high			*			Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56-62, 2005
					3380	c.2265C	3380C >A											
0370	Prostate cancer	Substitut	5 LBD		756	2266	p.(Thr756 Ala)						Somatic mutation		Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
					3381	c.2266A	3381A >G											
0602	Prostate cancer	Substitut	5 LBD		757	2269	p.(Asn757 Asp)						Flutamide treated - somatic mutation		Male	Normal		Taplin et al. J Clinical Oncology 21: 2673-2678, 2003
					3384	c.2269A	3384A >G											
0153	PAIS	Substitut	5 LBD		757	2270	p.(Asn757 Ser)								Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
					3385	c.2270A	3385A >G											
0532	MAIS	Substitut	5 LBD	*	757	2270	p.(Asn757 Ser)		high				Severe oligospermia-transactivation 38% of wt.		Male	Normal		Giwereman et al. Clin Endocrinol 54: 827-834, 2001
					3385	c.2270A	3385A >G											
1039	PAIS	Substitut	5 LBD		757	2270	p.(Asn757 Ser)						External Masculinisation Score = 12		Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
					3385	c.2270A	3385A >G											
1040	PAIS	Substitut	5 LBD		757	2270	p.(Asn757 Ser)						External Masculinisation Score = 12		Male	Ambiguous	pos	Hellmann et al. Arch Dis Child 97:403-409, 2012
					3385	c.2270A	3385A >G											
0573	Prostate cancer	Substitut	5 LBD		758	2272	p.(Val758 Ile)						Somatic mutation + Orchiectomy Horm-refractor y CaP		Male	Normal		Hyytinen et al. Lab Invest. 82: 1591-1598, 2002
					3387	c.2272G	3387G >A											
0300	Prostate cancer	Substitut	5 LBD	*	758	2273	p.(Val758 Ala)						Binds R1881 norm.- transcriptionally inactive- Som mut		Male	Normal		James et al. 79th US Endo Soc Meeting, Abstr P2-484, 1997
					3388	c.2273T	3388T >C											
0493	Prostate cancer	Substitut	5 LBD		758	2273	p.(Val758 Ala)						Somatic mutation		Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
					3388	c.2273T	3388T >C											
0346	PAIS	Substitut	5 LBD	*	759	2276	p.(Asn759 Thr)		normal		high	*	50% reduction in transactivation in COS-7					Yong et al. Mol & Cell Endocrinol. 137: 41-50, 1998
					3391	c.2276A	3391A >C											
0371	Prostate cancer	Substitut	5 LBD		760	2278	p.(Ser760 Pro)						Somatic mutation		Male	Normal		Takahashi et al. Cancer Research 55: 1621-1624, 1995
					3393	c.2278T	3393T >C											
0154	CAIS	Substitut	5 LBD		760	2279	p.(Ser760 Phe)		zero						Female	Normal		DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
					3394	c.2279C	3394C >T											
1092	CAIS	Substitut	5 LBD		760	2279	p.(Ser760 Tyr)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
					3394	c.2279C	3394C >A											
0605	CAIS	Deletion/insertion	5 LBD		761		p.(Arg761 fs)		zero				also 3396_3402del7 & 3408_3418ins11 stop 9 codon downstr.		Female	Normal	pos	Vichlis et al. J Hum Genet 48: 346-351, 2003

Accession #	Phenotype	Mutation type	Exon Domain	Pathogenicity proven CpG spot	Amino acid Base	Position HGVS nt	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0841	PAIS	Substitut	5 LBD		761		p.(Arg761 Ser)											Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0938	Prostate cancer	Substitut	5 LBD		761		p.(Arg761 )						Silent mut. Treated with anti-androgens. Occurred in >2 cases	Male	Normal			Steinkamp et al. Cancer Res 69: 4434-4442, 2009
0729	PAIS	Substitut	5 LBD		762	2285	p.(Met762 Thr)						mother heterozygous	Male	Ambiguous	pos		Ledig et al: Horm Res 63: 263-269, 2005
0155	CAIS	Substitut	5 LBD		763	2287	p.(Leu763 Phe)		zero					Female	Normal			Brown et al. 74th US Endo Soc Meeting, Abstr 1506, 1992
0156	CAIS	Substitut	5 LBD	*	763	2287	p.(Leu763 Phe)		zero					Female	Normal			Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
0861	CAIS	Substitut	5 LBD		763	2287	p.(Leu763 Phe)						Diag at 7yr bilateral gonadectomy at 8yrs	Female	Normal	pos		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0862	CAIS	Substitut	5 LBD		763	2287	p.(Leu763 Phe)						Diagnosed at 1 wk	Female	Normal	pos		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0157	CAIS	Substitut	5 LBD		764	2290	p.(Tyr764 His)							Female	Normal			Quigley et al. Endocrin. Reviews, 16: 271, 1995
0159	PAIS	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)		low					Male	Ambiguous			Morono et al. Human Mutation 6: 152-162, 1995
0405	PAIS	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)							Male	Ambiguous			Batch et al. Arch Disease Child 68: 453, 1993
0484	PAIS	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)		normal high									Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0485	PAIS	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)		normal high									Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0636	PAIS	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)	22	low				Gynecomastia -testis located in scrotum	Male	Ambiguous	neg		Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0372	Prostate Cancer	Substitut	5 LBD		764	2291	p.(Tyr764 Cys)						Somatic mutation	Male	Normal			Takahashi et al. Cancer Research 55: 1621-1624, 1995
0158	PAIS	Substitut	5 LBD	*	764	2291	p.(Tyr764 Cys)	12	normal high				* PolyGln tract short ( only 12 repeats)	Male	Ambiguous	pos		McPhaul et al. J Clin Inv 87: 1413,1991; Batch&al Arc Dis Ch
0850	CAIS	Substitut	5 LBD		764		p.(Tyr764 *)						Diag at 1 wk-Bilateral gonadectomy at 3wks	Female	Normal			Cheikhelard et al. J Urol 180: 1496-1501, 2008
0160	CAIS	Substitut	5 LBD	*	765		p.(Phe765 Leu)		low	high				Female	Normal	neg		Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0161	CAIS	Substitut	5 LBD		765	2293	p.(Phe765 Leu)		zero					Female	Normal			Ris-Stalpers et al: Pediatric Res,36. 227-234, 1994
0162	CAIS	Substitut	5 LBD		765	2295	p.(Phe765 Leu)		low	normal				Female	Normal			Pinsky et al. Clin Inv Med, 15: 456, 1992
0163	CAIS	Substitut	5 LBD	**	766	2296	p.(Ala766 Thr)		zero					Female	Normal			Bevan et al. J Steroid Biochem Mole Biol 61: 19-26, 1997
0164	CAIS	Substitut	5 LBD	*	766	2296	p.(Ala766 Thr)		zero					Female	Normal			Merkabi et al.75th US Endo Soc Meeting Abstr 602, 1993
0165	CAIS	Substitut	5 LBD	*	766	2296	p.(Ala766 Thr)							Female	Normal			Sweet et al. Fertil Sterility 58: 703, 1992
0166	CAIS	Substitut	5 LBD	*	766	2296	p.(Ala766 Thr)		zero					Female	Normal			Hiort et al. Am J Med Genet. 63: 218-222, 1996
0311	CAIS	Substitut	5 LBD	*	766	2296	p.(Ala766 Thr)	27						Female	Normal			Ko et al. J Reprod. Med 42: 424- 427, 1997
0382	CAIS	Substitut	5 LBD	*	766	2296	p.(Ala766 Thr)							Female	Normal			Giwerzman et al. Human Genetics 103: 529-531, 1998

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile						
0454	CAIS	Substitut	5 LBD	*	766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0455	CAIS	Substitut	5 LBD	*	766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0456	CAIS	Substitut	5 LBD	*	766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A						Woolfian remnants present		Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0585	CAIS	Substitut	5 LBD		766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A						Reduced immunoreactive AR		Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0586	PAIS	Substitut	5 LBD		766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A						Reduced immunoreactive AR		Female	Ambiguous		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0730	CAIS	Substitut	5 LBD		766 3411	2296 c.2296G	p.(Ala766 Thr) 3411G >A								Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0520	PAIS	Substitut	5 LBD		766 3411	2296 c.2296G	p.(Ala766 Ser) 3411G >T											Chavez et al. Clin Genet 59: 185-188, 2001
0167	CAIS	Substitut	5 LBD		766 3412	2297 c.2297C	p.(Ala766 Val) 3412C >T	20	zero						Female	Normal		Pinsky et al, Clin Inv Med, 15: 456-472, 1992
0168	CAIS	Substitut	5 LBD	*	767 3414	2299 c.2299C	p.(Pro767 Ser) 3414C >T		low	high	high				Female	Normal	pos	Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0457	CAIS	Substitut	5 LBD		767 3414	2299 c.2299C	p.(Pro767 Ser) 3414C >T								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0681	CAIS	Substitut	5 LBD		767 3414	2299 c.2299C	p.(Pro767 Ala) 3414C >G						bilateral inguinal hernia		Female	Normal		Bouvattier et al. J Clin Endocrinol Metab 87: 29-32, 2002
0543	CAIS	Substitut	5 LBD		767 3414	2299 c.2299C	p.(Pro767 Ala) 3414C >G		normal	high			2 affected siblings variable phenotype		Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0611	CAIS	Substitut	5 LBD		767 3414	2299 c.2299C	p.(Pro767 Ala) 3414C >G						Twin sisters		Female	Normal	pos	Correa et al. 16th Meet. Latin Amer.Soc Ped Endo. Abstr: 26, 2003
0587	CAIS	Deletion	5 LBD		767 3415	2300 c.2300	p.(Pro767 fs) 3415 delC						1 nt del.-frameshift & stop, low immunoreact AR		Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0169	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Single nt deletion causing frameshift & stop in codon 808		Female	Normal	pos	Baldazzi et al. Hum Mol Genet 3: 1169-1170, 1994
0388	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Single nt deletion causing frameshift & stop in codon 808		Female	Normal		Chung et al. Molecules & Cells 8: 741-745, 1998
0458	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Int del-frameshit & stop in codon 808 Wolffian remnants		Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0459	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Int del-frameshit & stop in codon 808 Wolffian remnants		Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0561	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Int del frame shift & stop in codon 808 in 2 unrelat individs		Female	Normal		Soriano Guillén et al. An Esp Pediatr 56: 347-352, 2002
0588	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						1 nt del framshift & stop codon 808 no immunoreact AR		Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0853	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Int del framshift & stop in codon 808 Diag - 7yr Wolff rem		Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0953	CAIS	Deletion	5 LBD		767 3416	2301 c.2301	p.(Pro767 fs) 3416 delT						Int del framshift & stop in codon 808 Male gender identity		Female	Normal		T'Sjoen et al. Arch Sex Behav 40: 655-638, 2011
0665	PAIS	Substitut	5 LBD		767 3414	2299 c.2299C	p.(Pro767 Ser) 3414C >T						Somatic mosaicism		Female	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0170	CAIS	Substitut	5 LBD		768 3419	2304 c.2304T	p.(Asp768 Glu) 3419T >G		v	low					Female	Normal		Lobaccaro et al. Pediatr Res 33: Abstr. 115, 1993
0343	CAIS	Substitut	5 LBD		768 3419	2304 c.2304T	p.(Asp768 Glu) 3419T >G								Female	Normal		Melo et al. 80th US Endo Soc Meeting Abstr P2-44, 1998

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0893	CAIS	Substitut	5 LBD		768	2302	p.(Asp768 Tyr)							Bone size intermediate between male and female	Female	Normal		Taes et al. Bone 45: 392-397, 2009
0544	PAIS	Substitut	5 LBD		769	2305	p.(Leu 769 Met)					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		769	2305	p.(Leu769 Val)				20			Testis located in labia majora	Female	Normal		Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0460	CAIS	Substitut	5 LBD		769	2306	p.(Leu769 Pro)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0666	CAIS	Deletion/Insertion	5 LBD		771	2312	p.(Phe771 *)							Somatic mosaicism sister heterozygous mother som mosaic ?	Female	Normal	pos	Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0171	PAIS	Substitut	5 LBD		772	2314	p.(Asn772 His)								Female	Ambiguous		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
0526	PAIS	Substitut	5 LBD	*	772	2314	p.(Asn772 His)					high		Size & level of expression of AR normal	Female	Ambiguous		Zhu et al, 83rd US Endo Soc Meeting, Abstr P2-34, 2001
0173	PAIS	Substitut	5 LBD		773	2318	p.(Glu773 Gly)					low high			Female	Ambiguous		Tincello et al. Clinical Endocrinology 46: 497-506, 1997
0174	PAIS	Substitut	5 LBD	*	773	2318	p.(Glu773 Ala)				25 23	normal normal high			Male	Ambiguous		Shkolny et al. J Clin Endocrinol Metab 84: 805-810, 1999
0172	CAIS	Substitut	5 LBD		773	2317	p.(Glu773 *)					zero			Female	Normal		Imasaki et al. Endocrine Journal 42: 643-648 1995
0839	CAIS	Deletion	5 LBD		773	2317	p.(Glu773 fs)							4 nt deletion leading to stop in codon 788	Female	Normal		Jeske et al. J Pediatr Endocrinol Metab 20: 893-908, 2007
0336	CAIS	Substitut	6 LBD	**	775	2323	p.(Arg775 Cys)				26 23	normal normal			Female	Normal		Prior et al. Am J Hum Genet 51: 143, 1992
0176	CAIS	Substitut	6 LBD	**	775	2323	p.(Arg775 Cys)				27 19	zero			Female	Normal	pos	Prior et al. Am J Hum Genet 51: 143, 1992
0177	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)					zero			Female	Normal		Mebarki et al. 72nd US Endo Soc Meeting, Abstr 791, 1990
0178	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)								Female	Normal		Hiort et al. J Pediatrics 132: 939-943, 1998
0179	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)					v low high			Female	Normal	neg	Marcelli et al. J Clin Endocrinol Metab 73: 318, 1991
0180	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)								Female	Normal		Jakubiczka et al. Human Mutation 9: 57-61, 1997
0331	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)								Female	Normal		Komori et al. Arch Gynecol & Obstetrics 261: 95-100, 1998
0175	CAIS	Substitut	6 LBD	**	775	2323	p.(Arg775 Cys)					v low			Female	Normal		Brown et al. Mol Endocrinol, 4: 1759-72, 1990
0355	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)							mosaic-de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998
0589	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)							Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0599	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)								Female	Normal		Scheiber et al. J Pedatric Endocrinol & Metab. 16: 367-373,
0731	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)							sister affected	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005
0732	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)							mother heterozygous	Female	Normal	pos	Ledig et al: Horm Res 63: 263-269, 2005
0182	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)					low normal	*		Female	Normal		Batch et al. Hum Mol Genet, 1: 497, 1992

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0183	CAIS	Substitut	6 LBD	**	775	2324	p.(Arg775 His)				v	low	high		Female	Normal		DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992
0184	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)								Female	Normal		Hiort et al. Am J Med Genet. 63. 218-222, 1996
0461	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)				zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0462	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0181	CAIS	Substitut	6 LBD	**	775	2324	p.(Arg775 His)				normal	high	high	* mutation found in two unrelated families	Female	Normal	pos	Prior et al. Am J Hum genet, 51: 143, 1992
0863	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)							Prenat diagnosis bilateral gonadectomy at 5yrs	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0878	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)								Female	Normal	pos	Qin et al. Zhonghua Fu Chan Ke Za Za 43: 828-830, 2008
0986	CAIS	Substitut	6 LBD	*	775	2323	p.(Arg775 Cys)	27	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0987	CAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)	20	17						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0185	PAIS	Substitut	6 LBD	*	775	2324	p.(Arg775 His)											Quigley et al. Endocrin Reviews 16: 271, 1995
0186	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)								Female	Normal		Hiort et al. Hum Mol Genet 3: 1163-1166 1994
0187	CAIS	Substitut	6 LBD	**	780	2338	p.(Arg780 Trp)								Female	Normal		Morono et al. Human Mutation 6: 152-162, 1995
0188	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)								Female	Normal		Sinnecker et al. Eur J. Pediatr. 156: 7-14, 1997
0463	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0621	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)	21			low			AR partially purified appears truncated (43 kD)	Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
0638	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)	24						Testis located in inguinal region	Female	Normal	neg	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0733	CAIS	Substitut	6 LBD	*	780	2338	p.(Arg780 Trp)								Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0189	PAIS	Substitut	6 LBD	*	781	2343	p.(Met781 Ile)				normal	high		*	Female	Ambiguous		Bevan et al. Hum Mol Genet, 5: 265-273, 1996
0190	PAIS	Substitut	6 LBD		781	2343	p.(Met781 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
0191	PAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)											Brinkmann et al. J Steroid Biochem & Mol Biol 53: 443, 1995
0192	PAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)							A brother to mutation 0305	Male	Ambiguous	pos	Rodien et al. J Clin Endo & Metab 81: 2904-2908, 1996
0305	CAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)							2 sisters to mutation 0192	Female	Normal	pos	Rodien et al. J Clin Endo & Metab 81: 2904-2908, 1996
0193	CAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)								Female	Normal		Jakubiczka et al. Human Mutation 9: 57-61, 1997
0464	CAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)				low	high		Epididymis & Vas deferens present - Cousin 660	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0660	CAIS	Substitut	6 LBD		781	2343	p.(Met781 Ile)							Epididymis & Vas deferens present -Cousin 464	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004



Accession #	Phenotype	Mutation type	Exon Domain	CpG spot	Pathogenicity proven	Amino acid Base	Position HGVS nt	Change Amino acid Base	Exon 1 tracts			Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
									Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile						
0198	MAIS	Substitut	6 LBD	794		2382	p.(Glu794 Asp)				normal	normal		Inconsistent increases in gynocomastia & infertility	Male	Normal		Pinsky et al. Clin Inv Med, 15: 456, 1992	
0397	Normal	Substitut	6 LBD	794		2382	p.(Glu794 Asp)							Homsosexual individual	Male	Normal		Macke et al. Am J Human Genetics 53: 844-852, 1993	
0199	CAIS	Substitut	6 LBD	795		2384	p.(Phe795 Ser)				zero				Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996	
0200	CAIS	Substitut	6 LBD	795		2384	p.(Phe795 Ser)								Female	Normal		Jakubiczka et al Human Mutation 9: 57-61, 1997	
0907	PAIS	Substitut	6 LBD	796		2388	p.(Gly796 Gly)				low	high		Silent mutation	Male	Ambiguous		Appari et al. J Mol Med 87: 623-632, 2009	
0201	CAIS	Substitut	6 LBD	* 797		2391	p.(Trp797 *)				v low				Female	Normal		Marcelli et al. J Clin Invest 85: 1522-1528, 1990	
0202	PAIS	Substitut	6 LBD	* 799		2395	p.(Gln799 Glu)				normal	normal	*		Female	Ambiguous		Bevan et al. Hum Mol Genet, 5: 265-273, 1996	
0203	PAIS	Substitut	6 LBD	799		2395	p.(Gln799 Glu)				normal	normal						Quigley et al. Endocrine Reviews 16: 271, 1995	
0204	PAIS	Substitut	6 LBD	799		2395	p.(Gln799 Glu)								Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996	
0205	Prostate cancer	Substitut	6 LBD	799		2395	p.(Gln799 Glu)							Also present in genomic DNA	Male	Normal		Evans et al. Prostate 28: 162-171, 1996	
0399	Prostate cancer	Substitut	6 LBD	799		2395	p.(Gln799 Glu)							Somatic mutation Stage 4 tumor	Male	Normal		Castagnaro et al. Verh. Dtsch. Ges. Path. 77. 119-123, 1993	
0340	MAIS	Substitut	6 LBD	* 799		2395	p.(Gln799 Glu)				normal			Azospemia	Male	Normal		Hiort et al. J Clin Endocrinol Metab 85: 2810-2815, 2000	
0381	MAIS	Substitut	6 LBD	* 799		2395	p.(Gln799 Glu)				normal			Azoospermia - defective transactivation	Male	Normal		Wang et al. J Clin Endocrinol Metab 83: 4303-4309, 1998	
0775	MAIS	Substitut	6 LBD	799		2395	p.(Gln799 Glu)							Male infertility	Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006	
0954	PAIS	Substitut	6 LBD	* 799		2395	p.(Gln799 Glu)							Virilization post-gonadectomy heteroz P450 (POR) p.Y601C	Female	Ambiguous		Idkowiak et al. J Clin Endocrinol Metab 95: 3418-3527, 2010	
1041	PAIS	Substitut	6 LBD	* 799		2395	p.(Gln799 Glu)							External Masculisation Score = 8	Male	Ambiguous		Hellmann et al. Arch Dis Child 97:403-409, 2012	
0648	CAIS	Substitut	6 LBD	799		2395	p.(Gln799 *)							Abdominal tumor - Sertoli cell adenoma	Female	Normal		Ignacek et al: Gynecol Endocrinol 19: 178-181, 2004	
0764	Prostate cancer	Substitut	6 LBD	800		2399	p.(Ile800 Thr)							patient lower Gleason score than patient -wt AR- som mutation	Male	Normal		Sanchez et al. BJU Int 98: 1320-1325, 2006	
0596	PAIS	Substitut	6 LBD	801		2402	p.(Thr801 Ile)							Variable expressivity- four additional males with MAIS	Male	Ambiguous	pos	Galli-Tsinopoulou et al. J Pediatr Endocrinol Metab16: 149-154,	
0542	CAIS	Deletion	6 LBD	801		2403	p.(Thr801 fs)							Single nt del causing frameshift & stop in codon 808 1 affect sib	Female	Normal	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001	
1055	CAIS	Deletion	6 LBD	801		2403	p.(Thr801 fs)							Single nt del causing frameshift & stop in codon 808 1 affect sib	Female	Normal	pos	Vasu et al. Genet Test Mol Biomarkers 16:749-755, 2012	
0521	PAIS	Substitut	6 LBD	803		2408	p.(Gln803 Arg)											Chavez et al. Clin Genet 59: : 185-188, 2001	
0498	CAIS	Substitut	6 LBD	804		2410	p.(Glu804 Lys)				zero	zero			Female	Normal	pos	Sawai et al. J Hum Genet 45: 342-345, 2000	
0563	CAIS	Substitut	6 LBD	805		2413	p.(Phe805 Leu)							de novo mutation. + glaucoma & hypertrophic pyloric stenosis	Female	Normal	neg	Gad et al. Clin Genet. 63: 59-63, 2003	
0865	CAIS	Substitut	6 LBD	805		2413	p.(Phe805 Ile)							Diagnosis at 1mo	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008	





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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0875	PAIS	Substitut	7 LBD	*	827 2481 3596 c.2481C	p.(Phe827 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	
0537	CAIS	Substitut	7 LBD		828 2482 3597 c.2482T	p.(Phe828 Val)								Female	Normal		Chavez et al. J Hum Genet. 46: 560-565, 2001	
1094	CAIS	Substitut	7 LBD		828 2483 3598 c.2483T	p.(Phe828 Ser)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012	
0622	CAIS	Insertion	7 LBD		830 2478 3593 c.2478_2479	p.(Glu830 fs)	18						2nt insertion causing frameshift and stop in Codon 834	Female	Normal	pos	MacLean et al. Hum Mutat. 23: 287, 2004	
0522	CAIS	Substitut	7 LBD		831 2491 3606 c.2494C	p.(Leu831 Val)								Female	Normal		Chavez et al. Clin Genet 59: : 185-188, 2001	
0213	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)			zero					Female	Normal	pos	DeBellis et al. Mol Endocrinol, 6: 1909-20, 1992	
0214	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)			zero					Female	Normal		Tincello et al. J Endocrinol, 132 Suppl, Abstr 87, 1992	
0215	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)			zero					Female	Normal		Ris-Stalpers et al. 74th Endo Soc Meeting, 1992	
0384	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)								Female	Normal		Giwereman et al. Human Genetics 103: 529-531, 1998	
0465	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)							Wolffian remnants present	Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004	
0500	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)								Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000	
0515	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)							Harmatoma found in pubertal patient	Female	Normal		Chen et al. Fertility & Sterility 74: 182-183, 2000	
0466	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)								Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000	
0499	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)								Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000	
0814	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)							"hamatomatous testes" present	Female	Normal		Goulis et al. Hirmines (Athens) 5: 200-204, 2006	
0216	CAIS	Substitut	7 LBD	**	832 2495 3610 c.2495G	p.(Arg832 Gln)			v low					Female	Normal	pos	Brown et al. Mol Endocrinol, 4: 1759-72, 1990	
0217	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)			zero				Found in two unrelated families	Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992	
0404	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)			zero					Female	Normal		Yaegashi et al. Tohoku J of Exp Med 187: 263-272, 1999	
0524	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)			zero				Sertoli cell carcinoma	Female	Normal		Ko et al. Int. J. Gynecol. Pathol. 20: 196-199, 2001	
0994	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Gln)	23	17					Mother heterozygous carrier	Female	Normal		Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0218	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Leu)	21	19	zero					Female	Normal		Shkolny et al. Human Mol Genetics 4: 515-521, 1995	
0307	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Leu)	26	16	zero					Female	Normal		Shkolny et al. Human Mol Genetics 4: 515-521, 1995	
0590	CAIS	Substitut	7 LBD	*	832 2495 3610 c.2495G	p.(Arg832 Leu)							Reduced immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002	
0995	CAIS	Substitut	7 LBD	*	832 2494 3609 c.2494C	p.(Arg832 *)	19	18						Female	Normal	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
1095	CAIS	Substitut	7 LBD			p.(Met833 *)								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012	



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									Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0229	PAIS	Substitut	7 LBD	*		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A				low							Imai et al. Annals of Clinical Biochem, 32: 482-486, 1995
0230	PAIS	Substitut	7 LBD	*		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A											Ghirri & Brown. Pediatr Res 33: Abstr.95, 1993
0231	PAIS	Substitut	7 LBD	**		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A				low	high	high					Marcelli et al. J Clin Invest 94: 1642-1650, 1994
0232	PAIS	Substitut	7 LBD	**		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A				normal	high		*	Female	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733 -
0223	PAIS	Substitut	7 LBD	**		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A				low	high			Female	Ambiguous		De Bellis et al. J Clin Endocrinol Metab, 78: 513, 1994
0668	PAIS	Substitut	7 LBD	*		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A							Somatic mosaicism	Male	Ambiguous		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005
0415	PAIS	Substitut	7 LBD			841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A	25						Testis located in inguinal region. same family as 0832	Male	Ambiguous		Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0626	PAIS	Substitut	7 LBD			841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A							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			841 3636	2521 c.2521C	p.(Arg841 Ser) 3636C >A							Testis located in inguinal region. same family as 0415	Male	Ambiguous		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
1008	PAIS	Substitut	7 LBD	*		841 3637	2522 c.2522G	p.(Arg841 His) 3637G >A	23	11					1 affected niece	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0233	PAIS	Substitut	7 LBD			842 3640	2525 c.2525T	p.(Ile842 Ser) 3640T >G				low	high			Female	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0234	CAIS	Substitut	7 LBD			843 3643	2528 c.2528T	p.(Ile843 Thr) 3643T >C				normal	high			Female	Normal	pos	Hiort et al. J Clin Endocrinol Metab, 77: 262-266, 1993
0235	PAIS	Substitut	7 LBD	*		843 3643	2528 c.2528T	p.(Ile843 Thr) 3643T >C				low	high		*	Male	Ambiguous	pos	Weidemann et al Clin Endocrinology 45: 733 - 739, 1996
0591	CAIS	Substitut	7 LBD			843 3643	2528 c.2528T	p.(Ile843 Asn) 3643T >A							Normal immunoreactive AR	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0736	PAIS	Substitut	7 LBD			844 3646	2531 c.2531C	p.(Ala844 Glu) 3646C >A							de novo mutation	Male	Ambiguous	neg	Ledig et al. Horm Res 63: 263-269, 2005
0494	Prostate cancer	Substitut	7 LBD			847 3654	2539 c.2539A	p.(Arg847 Gly) 3654A >G							Somatic mutation	Male	Normal		Marcelli et al. Cancer Research 60: 944-949, 2000
0236	CAIS	Insertion	7 LBD			849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA			zero				1 nt insert frameshift & stop in codon 880 & loss of 44 AA's	Female	Normal		Brinkmann et al. J Steroid Biochem Mol Biol 53: 443, 1995
0467	CAIS	Insertion	7 LBD			849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA			zero				1 nt insert causes frameshift.	Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0854	CAIS	Insertion	7 LBD			849		p.(Asn849 fs)							Frameshift & stop in Codon 880 Diag & bilat gonadect at 7y	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0957	CAIS	Insertion	7 LBD			849 3661	2546 c.2546_2547	p.(Asn849 fs) 3661_3662 insA							1 nt insert - frameshift & stop in Codon 880	Female	Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010
1096	CAIS	Substitut	7 LBD			849 3663	2548 c.2548T	p.(Asn Lys) 3663T								Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0237	CAIS	Substitut	7 LBD			854 3676	2561 c.2561C	p.(Ser854 *) 3676C >G				zero				Female	Normal		Wilson et al. J Clin Endocrinol Metab, 75: 1474-8, 1992
0238	CAIS	Substitut	7 LBD			854 3676	2561 c.2561C	p.(Ser854 *) 3676C >G				zero				Female	Normal		Jakubiczka et al. Human Mutation 9: 57-61, 1997
0793	CAIS	Deletion	7 LBD			854 3677	2562 c.2562_2563	p.(Ser854 ) 3677_3678 delA							2 nt deletion	Female	Normal		Mueller et al. Hum Genet 119: 673, 2006
0239	PAIS	Substitut	7 LBD			855 3679	2564 c.2564G	p.(Arg855 Lys) 3679G >A				low			*				McPhaul et al. J Clin Inv, 90: 2097, 1992

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								Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
1056	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T								Female	Normal		Vasu et al. Genet Test Mol Biomarkers 16:749-755, 2012
0240	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				zero				Female	Normal		DeBellis et al. Mol Endocrinol 6: 1909-20, 1992
0241	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T								Female	Normal		Tincello et al. J Endocrinol 132 Suppl, Abstr 87, 1992
0242	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				zero				Female	Normal		McPhaul et al. J Clin Inv, 90: 2097, 1992
0243	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T								Female	Normal		Loboccaro et al. Pediat Res 33: Abstr 115, 1993
0244	CAIS	Substitut	7 LBD	**	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				low				Female	Normal	pos	Morono et al. Human Mutation 6: 152-162, 1995
0245	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				zero				Female	Normal		Sultan et al. J Steroid Biochem & Mol Biol: 40
0246	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T								Female	Normal		Brinkmann et al. J Steroid Biochem & Mol Biol 53: 443, 1995
0247	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				zero				Female	Normal		Hiort et al. Am J Med Genet. 63: 218-222, 1996
0248	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				v low	high			Female	Normal	pos	Malmgren et al. Clin Genet. 50: 202-205, 1996
0320	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T								Female	Normal		Komori et al: J Obstetrics & Gynecol. Res. 23: 277-81, 1997
0468	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				zero				Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0469	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				normal	high			Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0527	CAIS	Substitut	7 LBD	**	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T				v low	high			Female	Normal		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
0592	CAIS	Substitut	7 LBD	**	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						v low immunoreactive AR		Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002
0640	CAIS	Substitut	7 LBD	**	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T	19			zero		Testis located in abdomen Same family as 0833		Female	Normal	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0737	CAIS	Substitut	7 LBD	*	856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						de novo mutation		Female	Normal	neg	Ledig et al: Horm Res 63: 263-269, 2005
0661	CAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						Vas deferens present		Female	Normal		Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0528	PAIS	Substitut	7 LBD	**	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A				normal	high		*	Male	Ambiguous		Elhaji et al. 83rd US Endo Soc Meeting, Abstr P2-37, 2001
0684	CAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						bilateral inguinal hernia		Female	Normal		Bouvattier et al: J Clin Endocrinol & Metab 87: 29-32, 2002
0688	CAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						Mother a heterozyote		Female	Normal		Skordis et al. J Pediatr Endocrinol Metab 18: 309-313, 2005
0745	PAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						sibling of 0746		Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0746	PAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A				normal	high		niece of 0745 also P392S	Female	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001
0777	MAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						Male infertility		Male	Normal		Ferlin et al. Clin Endocrinol 65: 606-610, 2006
0641	PAIS	Substitut	7 LBD	*	856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	19			zero to normal		Testis located in inguinal region		Male	Ambiguous	neg	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003

Accession #	Phenotype	Mutation type	Exon Domain	Pathogenicity proven CpG hot spot	Amino acid Base	Position HGVS nt base	Change Amino acid Base	Exon 1 tracts Poly Gln #	Poly Gly #	Androgen Binding			Comments	Sex of rearing	External Genitalia	Family history	Reference
										Bmax	Kd	Thermolabile k					
0716	PAIS	Substitut	7 LBD		856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					high		Male	Ambiguous		Deeb et al. Clinical Endocrinology 63: 56-62, 2005
0251	PAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					normal high					Chang et al. 73rd Endo Soc Meeting, Abstr 28, 1991
0252	PAIS	Substitut	7 LBD		**856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					normal high	* Servere hypospadi	Male	Ambiguous	pos	Batch et al. Hum Mol Genet, 1: 497, 1992
0253	PAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A							Male	Ambiguous		Hiort et al. Am J Med Genet. 63: 218-222. 1996
0254	PAIS	Substitut	7 LBD		**856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					zero		Female	Ambiguous	pos	Weidemann et al. Clin Endocrinology 45: 733-
0255	PAIS	Substitut	7 LBD		**856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					low high norm		Female	Ambiguous		Marcelli et al. J Clin Invest, 94: 1642-1650, 1994
0301	PAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	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		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					zero		Female	Ambiguous		Weidemann et al. Clin Endocrinology 45: 733-739, 1996
0302	PAIS	Substitut	7 LBD		**856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	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		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A					low		Female	Normal		McPhaul et al. J Clin Invest. 90: 2097, 1992
0344	PAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	28				zero to normal	Sex changed at 1yr Male to female - Same family as 0834	Female	Ambiguous	pos	Melo et al. J Clin Endocrinol & Metab 88: 3241-3250, 2003
0833	CAIS	Substitut	7 LBD		*856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						gonads located in abdomen. Same family as 0604	Female	Normal		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0834	PAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A						Same family as 0344	Male	Ambiguous	pos	Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005
0866	CAIS	Substitut	7 LBD		*856 3681	2566 c.2566C	p.(Arg856 Cys) 3681C >T						Diagnosis at 3 wks	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
0998	CAIS	Substitut	7 LBD		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	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		*856 3682	2567 c.2567G	p.(Arg856 His) 3682G >A	18	17				1 affected sister, mot & 2 aunts heterozygote carriers	Female	Ambiguous	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
0470	CAIS	Substitut	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000
0662	CAIS	Substitut	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G						Also S866P-twin of 0663	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0663	CAIS	Substitut	7 LBD		857 3686	2571 c.2571C	p.(Phe857 Leu) 3686C >G						Also S866P-twin of 0662	Female	Normal	pos	Hannema et al. J Clin Endocrinol Metab 89: 5815-5822, 2004
0356	CAIS	Substitut	7 LBD		858		p.(Tyr858 *)						de novo mutation	Female	Normal	neg	Hiort et al. J Pediatrics 132: 939-943, 1998
0941	CAIS	Duplicat	7 LBD		858 3688	2573 c.2573	p.(Tyr858 *) 3688 dupA						Int duplicatn resulted in stop codon. Mother & sister carriers	Female	Normal	pos	Turek-Plewa et al. Hum Genet 125: 341, 2009
0753	CAIS	Substitut	7 LBD		*860 3693	2578 c.2578C	p.(Leu860 Phe) 3693C >T					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		861 3697	2582 c.2582C	p.(Thr861 Asn) 3697C >A						Diagnosis at birth bilateral gonadectomy at 15yrs	Female	Normal		Cheikhelard et al. J Urol 180: 1496-1501, 2008
1097	CAIS	Substitut	7 LBD		861 3697	2582 c.2582C	p.(Thr861 Ile) 3697C >T							Female	Normal		Hughes et al. Semin Reprod Med 30: 432-442, 2012
0256	CAIS	Substitut	7 LBD		864 3706	2591 c.2591T	p.(Leu864 Arg) 3706T >G							Female	Normal		Brown et al. Eur J Pediatr 152: (Suppl 2) S62, 1993



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									Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile k						
0855	CAIS		7 LBD			867		p.(Val867 fs)							Frameshift & stop in Codon 876 Diag 15yr Bilat gonadect at 16y	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0856	CAIS		7 LBD			867		p.(Val867 fs)							Frameshift & stop in Codon 876 Diag 11yr Bilat gonadect at 11yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0259	PAIS	Substitut	7 LBD			867	2599 3714 c.2599G	p.(Val867 Leu) 3714G >T	21			normal high				Male	Ambiguous	pos	Saunders et al. Clin Endocrinol 37: 214, 1992
0851	CAIS	Substitut	7 LBD			868	2602 3717 c.2602C	p.(Gln868 *) 3717C >T							Diag at 2.5 yr	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0852	CAIS	Substitut	7 LBD			868	2602 3717 c.2602C	p.(Gln868 *) 3717C >T							Diag at 2 wk	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0940	Prostate Cancer	Substitut	7 LBD			868	2602 3717 c.2602C	p.(Gln868 *) 3717C >T							Both treated and untreated Occurred in 2 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009
1054	Female Breast Cancer	Substitut	7 LBD			868	2604 3719 c.2604G	p.(Gln868 His) 3719G >T							MDA-MB-453 breast cancer cell line reduced sensi to DHT	Female	Normal		Moore et al. Endocr Related Cancer 19:599-613, 2012
0601	CAIS	Insertion	7 LBD			869		p.(Pro869 ) insA TG							3 base insertion (ATG) at codon 869	Female	Normal		Scheiber et al. J Pediatric Endo Metab 16: 367-373, 2003
0269	PAIS	Substitut	8 LBD	*		870	2610 3725 c.2610T	p.(Ile870 Met) 3725T >G				normal high	*	Hypospadias	Male	Ambiguous	pos	Bevan et al. Hum Mol Genet 5: 265-273, 1996	
0718	PAIS	Substitut	8 LBD			870	2610 3725 c.2610T	p.(Ile870 Met) 3725T >G				high				Male	Ambiguous	pos	Deeb et al. Clinical Endocrinology 63: 56-62, 2005
0270	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Val) 3727C <T							Found in two unrelated families	Male	Ambiguous		Hiort et al. Eur J Pediatr, 153: 317, 1994
0315	PAIS	Substitut	8 LBD			871	2612 3727 c.2612C	p.(Ala871 Gly) 3727C >G							Severe hypospadias	Male	Ambiguous		Albers et al. J of Pediatrics 131: 388-392, 1997
0271	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Gly) 3727C >G							de novo mutation	Female	Ambiguous	neg	Hiort et al. J Pediatrics 132: 939- 943, 1998
0562	MAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Gly) 3727C >G							bilateral gynecomastia normal fertility	Male	Normal		Zenteno et al. Horm Res 57: 90-93, 2002
0794	CAIS ?	Deletion	8 LBD			871	2613 3728 c.2613	p.(Ala871 ) 3728 delG							1 nt deletion				Mueller et al. Hum Genet 119: 673, 2006
0951	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Val) 3727C >T							Micropenis only-Hetrozygous for SRD5A2 V89L	Male	Ambiguous		Bhangoo et al. Asian J Androl 12: 561-566, 2010
1010	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Val) 3727C >T	25	11						Male	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010
1098	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Val) 3727C >T											Hughes et al. Semin Reprod Med 30: 432-442, 2012
1099	PAIS	Substitut	8 LBD	*		871	2612 3727 c.2612C	p.(Ala871 Glu) 3727C >A											Hughes et al. Semin Reprod Med 30: 432-442, 2012
0272	MAIS	Substitut	8 LBD	*		872	2614 3729 c.2614A	p.(Arg872 Gly) 3729A >G	26	24		normal normal norm			Gynocomastia & oligospermia	Male	Normal		Shkolny et al. J Clin Endocrinol Metab 84: 805-810, 1999
0696	Prostate cancer	Substitut	8 LBD	*		873	2617 3732 c.2617G	p.(Glu873 Gln) 3732G >C	24			normal normal			Activated by estradiol, progesterone & CPA	Male	Normal		Chen et al. The Prostate 63: 395-406, 2005
0273	Prostate cancer	Substitut	8 LBD			875	2623 3738 c.2623C	p.(His875 Tyr) 3738C >T							Som mut- stimulated by progesterone & oestrogen	Male	Normal		Taplin et al. New England J Med 332: 1393-1398, 1995
0274	Prostate cancer	Substitut	8 LBD			875	2623 3738 c.2623C	p.(His875 Tyr) 3738C >T							Somatic mutation	Male	Normal		Tan et al. J of Urology 155: 340A, 1996
0538	CAIS	Substitut	8 LBD			875	2624 3739 c.2624A	p.(His875 Arg) 3739A >G				zero				Female	Normal		Chavez et al. J Hum Genet. 46: 560-565, 2001
0868	CAIS	Substitut	8 LBD			875	2624 3739 c.2624A	p.(His875 Arg) 3739A >G							Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008



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								Poly Gln #	Poly Gly #	Bmax	Kd	k						
0869	CAIS	Substitut	8 LBD		875	2624	p.(His875 Arg)							Prenatal diagnosis	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0870	CAIS	Substitut	8 LBD		875	2624	p.(His875 Arg)							Diagnosis at 2mo bilateral gnadectomy at 16yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0871	CAIS	Substitut	8 LBD		875	2624	p.(His875 Arg)							Diagnosis at 1yr bilateral gonadectomy at 15yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0872	CAIS	Substitut	8 LBD		875	2624	p.(His875 Arg)							Diagnosis at 7 yrs bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0873	CAIS	Substitut	8 LBD		875	2624	p.(His875 Arg)							Prenatal diagnosis & bilateral gonadectomy at 14yrs	Female	Normal	pos	Cheikhelard et al. J Urol 180: 1496-1501, 2008
0275	LNCaP cell line	Substitut	8 LBD		878	2632	p.(Thr878 Ala)							Altered binding specificity - somatic mutation	Male	Normal		Veldscholte et al. Biochem Biophys Res Comm, 172: 534, 1990
0276	Prostate cancer	Substitut	8 LBD		878	2632	p.(Thr878 Ala)							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		878	2632	p.(Thr878 Ala)							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		878	2632	p.(Thr878 Ala)							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		878	2632	p.(Thr878 Ala)							Somatic mutation in bone metastases of Prostate cancer	Male	Normal		Kleinerman et al. J of Urology 155: 624A, 1996
0432	Prostate cancer	Substitut	8 LBD		878	2632	p.(Thr878 Ala)							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		878	2632	p.(Thr878 Ala)							Somatic mutation - flutamide treated	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
0604	Prostate cancer	Substitut	8 LBD		878	2632	p.(Thr878 Ala)							Somatic mutation - flutamide treated	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
0647	Prostate cancer	Substitut	8 LBD	*	878	2632	p.(Thr878 Ala)							+Q641*- late stage disease- som mut - transactivates	Male	Normal		Ceraline et al. Intl J Cancer 108: 152-157, 2003
0280	Prostate cancer	Substitut	8 LBD	*	878	2633	p.(Thr878 Ser)							Som mut. in 86% of isolates. Stimulated by estrogen & progester	Male	Normal		Taplin et al: New England J Med 332: 1393-1398, 1995
1026	AIS	Substitut	8 LBD		878	2633	p.(Thr878 Ile)											Mueller et al. Hum Gen 127: 479, 2010
0539	PAIS	Substitut	8 LBD		880	2638	p.(Asp880 Tyr)				normal				Male	Ambiguous		Chavez et al. J Hum Genet. 46: 560-565, 2001
0553	Prostate cancer	Substitut	8 LBD		880	2639	p.(Asp880 Gly)							Treated with bicalumotide - somatic mutatation	Male	Normal		Taplin et al. J Clinical Oncology 21: 2673 -2678, 2003
0281	CAIS	Substitut	8 LBD		882	2644	p.(Leu882 Val)							Somatic instabilty in polyglutamine tract	Female	Normal	pos	Davies et al. Clinical Endocrinology 43: 69 -77, 1995
1100	PAIS	Substitut	8 LBD		882	2644	p.(Leu882 Ile)											Hughes et al. Semin Reprod Med 30: 432 -442, 2012
0829	CAIS	Substitut	8 LBD		882	2645	p.(Leu882 Pro)								Female	Normal	neg	Galani et al. Fertility & Sterility 2008
0282	CAIS	Substitut	8 LBD		884	2650	p.(Lys884 *)				zero				Female	Normal	pos	Trifiro et al. Am J Med Genet, 40: 493, 1991
0623	CAIS	Substitut	8 LBD		885	2654	p.(Ser885 *)				23	v low			Female	Normal		MacLean et al. Hum Mutat. 23: 287, 2004
0283	MAIS	Substitut	8 LBD	*	887	2659	p.(Met887 Val)				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	*	887	2659	p.(Met887 Val)				21	24	normal normal norm	Oligospermia-50% red. in transactivation	Male	Normal		Ghadessy et al. J. Clin. Endocrinol. 103: 1517 -1525, 1999





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									Poly Gln #	Poly Gly #	Bmax	Kd	Thermolabile						
0293	PAIS	Substitut	8 LBD	914		2740	p.(Pro914 Ser)	3855C >T										Ghirri and Brown. Paed Res. 33(5) Suppl, Abstr 95, 1993	
1015	PAIS	Substitut	8 LBD	914		2740	p.(Pro914 Ser)	3855C >T			zero	zero			Female	Ambiguous	neg	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0958	CAIS	Substitut	8 LBD	914		2741	p.(Pro914 Arg)	3856C >G							Female	Normal	neg	Wu et al. Fertility & Sterility 93: 2076, e1-4, 2010	
0844	CAIS	Deletion	8 LBD	* 916			p.(Phe917 *)						12nt del. Stop in codon 917 affect sister & aunts. neg. N/C interaction		Female	Normal	pos	Werner et al. Sex Dev 2: 73-83, 2008	
0318	CAIS	Substitut	8 LBD	* 917		2751	p.(Phe917 Leu)	3866C >G			low	high	*		Female	Normal		Radnayr et al. J of Urology 158: 1553-1556, 1997	
0477	CAIS	Substitut	8 LBD	918		2753	p.(His918 Arg)	3868A >G							Female	Normal		Ahmed et al. J Clin Endocrinol Metab 85: 658-665, 2000	
0303	Prostate cancer	Substitut		* 920		2759	p.(Gln920 Arg)	3874A >G					Somatic mutation		Male	Normal		Nazareth et al. 79th US Endo Soc Meetings Abstr. P2-498, 1997	
0294	CAIS	Splice	exon1/ intron 1			c.1616+2 1616+3	2731+2_2731 +3 insT			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			c.1616+5G	2731+5G >C						Substitution at +5 of donor splice site		Female	Normal	pos	Philibert et al. Fertility & Sterility 94: 472-476 2010	
0304	CAIS	Splice	exon2/ intron 2			c.1768+1G	2883+1G >A						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	
0351	CAIS	Splice	exon2/ intron 2			c.1768+1G	2883+1G >A						Substitution at +1 pos of donor splice site		Female	Normal		Hiort et al. J Pediatrics 132: 939- 943, 1998	
0317	Breast Cancer	Deletion	exon3										delta-exon3AR mRNA: higher express of mut. var in 7/13 breast cancer		Female	Normal		Zhu X et al. Int J Cancer 72: 574-580, 1997	
1022	Premature ovarian failure	Substitut	exon3/ intron 3			c.1885+9C	3000+9C >A						Substitution at +9 of donor site. Patient had menopause at 18		Female	Normal		Panda et al. Gynecol Endocrinol 27: 1-7, 2011	
0295	CAIS	Splice	exon3/ intron 3			c.1885+1G	3000+1G >A						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			c.1885+1G	3000+1G >A				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			c.2173+1G	3288+1G >T				zero		+1 pos of donor site. Splice site activated & p.(684G_724Pdel)		Female	Normal		Ris-Stalpers et al. Proc Natl AcadSci 87: 7866-70, 1990	
1057	CAIS	Splice	exon5/ intron 5			c.2320+1G	3435+1G >A						Mother + a sister carriers		Female	Normal	pos	Vasu et al. Genet Test Mol Biomarkers 16:749-755, 2012	
1018	CAIS	Splice	exon6/ intron 6			c.2450-44G	3565-44G >A			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
0503	PAIS	Splice	exon6/ intron 6			c.2449+5G	3564+5G >T				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			c.2449+5G	3564+5G >C				zero		Substit. at +5 position of donor splice site.		Female	Normal		Chavez et al. J Hum Genet 46: 560-565, 2001	
1029	CAIS	Splice	exon6/ intron 6			c.2449+3A	3564+3A >T						Substitution at +3 pos of donor splice site. Exon 6 skipped					Trifiro et al. Eur J Hum Genetics 5: 50-58, 1997	
0298	CAIS	Splice	exon7/ intron 7			c.2607+1G	3722+1G >A				zero		Substit. +1 pos donor splice, mRNA exon 7del, stop + 10 aa from exon 8		Female	Normal	pos	Lim et al. Mol & Cell Endocrinology 131: 205-210, 1997	
0502	CAIS	Splice	exon7/ intron 7			c.2607+1G	3722+1G >T						Substitution at + 1 position of donor splice site		Female	Normal		Choi et al. Arch Gynecol Obstet 263: 201-205, 2000	

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									Poly Gln #	Poly Gly #	Bmax	Kd	k	Thermolabile					
0670	CAIS	Splice	exon7/intron 7				c.2607+1G	3722+1G >A					normal normal	Substit. at + 1 pos of donor splice site somtaic mosaicism	Female	Normal		Kohler et al. J Clin Endocrinol Metab 90: 106-111, 2005	
0088	PAIS	Deletion	intron 2				c.?-1769-19	?_2884-19 del					normal normal	≥6 kb del at -19 pos of acceptor site. 2 transcr: wt, exon3del	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, affects splicing, related to 0750	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001	
0750	PAIS	Deletion	intron 2											related to 0749	Male	Ambiguous	pos	Boehmer et al. J Clin Endocrinol Metab 86: 4151-4160, 2001	
0299	PAIS	Splice	intron 2/exon 3				c.1769-11T	2884-11T >A						Subst. -11 slice accept. 2 transcr: exon3del & +69 nt; 3 affect sib, var phen	Male	Normal	pos	Bruggenwirth et al. Am J Hum Genet 61: 1067-1077, 1997	
0809	Prostate cancer	Splice	intron 2/exon 3	*			c.1768_1769	2883_2884 ins69						AR23 variant, ins 69 nt of intron 2 = 23 aa, affects AR trafficking	Male	Normal		Jagla et al. Endocrinology 148: 4334-43, 2007	
1017	CAIS	Splice	intron 2/exon 3				c.1769-3C	2884-3C >G			26	18		Substit at -3 of acceptor site. Aborted fetus	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0909	Prostate cancer	Splice	intron 2/exon 3				c.1768_1769	2883_2884 ins69						AR23 splice variant, ins 69 nt of intron 2, found in 5/8 cases	Male	Normal		Steinkamp et al. Cancer Res 69: 4434-4442, 2009	
0800	CAIS	Splice	intron 2/exon 3				c.1769-1G	2884-1G >A						Subst. -1 pos accept. transcr: 2883_2884ins69	Male	Normal		Jaaskelainen et al. Hum Mutat 27: 291, 2006	
1016	CAIS	Splice	intron 2/exon 3				c.1769-2A	2884-2A >C			13	18		Substit at -2 of acceptor site. Also Lys591Glu mut	Female	Normal	pos	Audi et al. J Clin Endocrinol Metab 95: 1876-1888, 2010	
0594	CAIS	Splice	intron 3/exon 4				c.1886-1G	3001-1G >T						Substit at -1 of acceptor site.	Female	Normal		Avila et al. J Clin Endocrinol Metab 87: 182-188, 2002	
0835	PAIS	Substitut	intron 3/exon 4				c.1886-60G	3001-60G >A						Male to female	Female	Ambiguous		Melo et al. Arq Bras Endocrinol Metab 49: 87-97, 2005	
0323	Prostate cancer		3'UTR											Polymorphism in 3'UTR Somatic mutation	Male	Normal		Paz et al. European Urology 63:56-62, 2005	