Table S1 Summary of *PITX2* intragenic mutations

Nucleotide change	Predicted protein change	Exon	Domain	Reference
c.47-1G>T	Splice site	IVS4		(Maciolek *et al.*, 2006)
c.47-1G>C	Splice site	IVS4		(Perveen *et al.*, 2000)
c.114del	p.Q39Kfs*116	5		(Strungaru *et al.*, 2007)
c.127C>T	p. R43W	5	HD	(Idrees *et al.*, 2006)
c.134dupA	p.H45Qfs*154	5	HD	(Reis *et al.*, 2012)
c.134_137del	p.H45Lfs*109	5	HD	(Wang *et al.*, 2003)
c.137_138del	p.F46Yfs*152	5	HD	(D'haene *et al.*, 2011)
c.143_144del	p.S48Tfs*150	5	HD	(Reis, *et al.*, 2012)
c.151_171dup	p.F58Lfs*222	5	HD	(Priston *et al.*, 2001)
c.160-252_253-734del	p.L55Qfs*46	5-6	HD and OAR	(De La Houssaye *et al.*, 2006)
c.161T>A	p.L54Q	5	HD	(Semina *et al.*, 1996)
c.163G>T	p.E55*	5	HD	(Vieira *et al.*, 2006)
c.172T>C	p.F58L	5	HD	(D'haene, *et al.*, 2011)
c.174C>G	p. F58L	5	HD	(Vieira, *et al.*, 2006)
c.175C>T	p.Q59*	5	HD	(D'haene, *et al.*, 2011)
c.185G>A	P.R62H	5	HD	(Amendt *et al.*, 2000)
c.191C>G	p.P64R	5	HD	(Weisschuh *et al.*, 2006)
c.191C>T	p.P64L	5	HD	(Phillips, 2002)
c.198_201delinsTTTCT	p.M66Ifs*133	5	HD	This study
c.199C>T[a]	p.Q67*	5	HD	(Law *et al.*, 2011)
c.202A>C	p.T68P	5	HD	(Semina, *et al.*, 1996)
c.206G>A	P.R69H	5	HD	(Kulak *et al.*, 1998)
c.224G>A	p.W75*	5	HD	(D'haene, *et al.*, 2011)
c.225G>A	p.W75*	5	HD	(Reis, *et al.*, 2012)
c.247G>C	p.V83L	5	HD	(Priston, *et al.*, 2001)
c.250C>T	p.R84W	5	HD	(Alward *et al.*, 1998)
c.252+5G>C	Splice site	IVS5		(Semina, *et al.*, 1996)
c.253-11A>G	Splice site	IVS5		(Semina, *et al.*, 1996)
c.253-2A>T	Splice site	IVS5		(Doward *et al.*, 1999)
c.253-1G>A	Splice site	IVS5		(Reis, *et al.*, 2012)
c.257G>C	p.W86S	6	HD	(Reis, *et al.*, 2012)
c.258G>T	P.W86C	6	HD	(Li *et al.*, 2008)
c.262A>G	p.K88E	6	HD	(Perveen, *et al.*, 2000)
c.268C>T	p.R90C	6	HD	(Perveen, *et al.*, 2000)
c.269G>C	p.R90P	6	HD	(Phillips, 2002)
c.272G>C	p.R91P	6	HD	(Semina, *et al.*, 1996)
c.282G>A	p.W94*	6	HD	(Amendt, *et al.*, 2000)
c.286_287del	p.K96Efs*102	6	HD	(Perveen, *et al.*, 2000)
c.289_290del	p.R97Gfs*101	6	HD	(D'haene, *et al.*, 2011)
c.301C>T	p.Q101*	6		(D'haene, *et al.*, 2011)
c.304C>T	p.Q102*	6		(D'haene, *et al.*, 2011)
c.313C>G	p.L105V	6		(Phillips, 2002)
c.323A>C	p.N108T	6		(Phillips, 2002)
c.356del	p.Q119Rfs*36	6		(Perveen, *et al.*, 2000)
c.363C>A	p.Y121*	6		(Vieira, *et al.*, 2006)
c.366del	p.D122Efs*33	6		(Saadi *et al.*, 2001)
c.398G>A	p.W133*	6		(Semina, *et al.*, 1996)
c.410G>T	p.G137V	6		(Kniestedt *et al.*, 2006)
c.416del	p.T139Nfs*16	6		(Strungaru, *et al.*, 2007)
c.500dup	p.P168Tfs*31	6		(Perveen, *et al.*, 2000)
c.652_653delinsAAG	p.Y218Qfs*11	6		(Perveen, *et al.*, 2000)
c.662_669dup	p.P224Rfs*18	6		(Vieira, *et al.*, 2006)
c.679del	p.Y227Mfs*12	6		(Brooks *et al.*, 2004)
c.690del	p.C231Vfs*8	6		(Borges *et al.*, 2002)
c.698C >T	p.S233L	6	OAR	(Kelberman *et al.*, 2011)
c.708_730del	p.S237Afs *48	6	OAR	(Reis, *et al.*, 2012)

[a] Numbering is relative to the coding DNA sequence of *PITX2B* (NM_153426), others are relative to the coding DNA sequence of *PITX2A* (NM_153427.1), where nucleotide +1 is the A of the ATG translation initiation codon. Mutation is named according to the HGVS Mutation Nomenclature Recommendations (http://www.hgvs.org/mutnomen/recs.html/provided in the public domain by the Human Genome Variation Society). HD, homeodomain; OAR, OAR domain

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