Table S1 Molecular Data

	Inheritance	FOXP1 mutation	Mutation type
Patient 1	dn	arr3p13(70949677-71366497)x1 dn	Partial deletion (Exon 5 - 21)
Patient 2	dn	c.1573C>T ; p.(R525*)	Nonsense mutation
Patient 3	NA	c.1526G>A ; p.(W509*)	Nonsense mutation
Patient 4	dn	arr3p13(71077030-71200157)x1 dn	Partial deletion (Exon 7 - 11)
Patient 5	dn	c.858delC ; p.(P286fs*38)	Frameshift
Patient 6	dn	c.1543C>G ; p.(H515D)	Missense mutation
Patient 7 (Decipher 331530)	dn	arr3p13(70734112-71097793)x1 dn	Partial deletion (Exon 10 - 21)
Patient 8	dn	c.1573C>T ; p.(R525*)	Nonsense mutation
Patient 9 (Decipher 255792)	dn	arr3p14.1p12.3(69317837-74319827)x1 dn	Whole deletion
Patient 10 (Decipher 260107)	dn	arr3p14p13(66604274-73581090)x1 dn	Whole deletion
Patient 11 (Decipher 272645)	dn	arr3p14.1p13(67891690-72013271)x1 dn	Whole deletion
Patient 12 (Decipher 274063)	dn	arr3p13(70569005-71199976)x1 dn	Partial deletion (Exon 7 - 21)
Patient 13 (Decipher 284583)	dn	arr3p13(71030149-71379636)x1 dn	Partial deletion (Exon 5 - 14)
Patient 14 (Decipher 272245)	dn	c.974+1G>C ; p.?	Splice site
Patient 15 (Decipher 274751)	dn	c.1579G>T ; p.(E527*)	Nonsense mutation
Patient 16 (Decipher 262793)	dn	c.1169C>T ; p.(T390I)	Missense mutation
Patient 17 (Decipher 279456)	dn	c.1468delCInsAACAC ; p.L490Nfs*4	Frameshift
Patient 18 (Decipher 270887)	dn	arr3p14.1p13(66668049-73248306)x1 dn	Whole deletion
Patient 19 (Decipher 252324)	dn	arr3p13p12.3(71366497-74373822)x1 dn	Partial deletion (Exon 1 - 4)
Patient 20	dn	c.1429-2A>G ; p.?	Splice site
Patient 21	NA	c.1556_1560delTTCAC ; p.(L519Qfs*12)	Frameshift
Patient 22	NA	c.511-1G>A ; p.?	Splice site
Patient 23	dn	c.1573C>T ; p.(R525*)	Nonsense mutation
Patient 24	dn	arr3p13p12.3(71213948-77496712)x1 dn	Partial deletion (Exon 1 - 6)
Patient 25	dn	c.1319C>G ; p.(S440*)	Nonsense mutation
PARIANI ET AL	dn	arr3p14.1p13(71115010 -71909694)x1 dn	Partial deletion (Exon 1 - 6)
HORN ET AL - Patient 1	NA	arr3p(70725077-71223275)x1	Partial deletion (Exon 6 - 21)
HORN ET AL - Patient 2	dn	arr3p(70695380-71354664)x1 dn	Partial deletion (Exon 4 - 21)
HORN ET AL - Patient 3	dn	arr3p(70258557-71305483)x1 dn	Partial deletion (Exon 5 - 21)
HAMDAN ET AL - Patient 1	dn	arr3p(71032185-71421950)x1 dn	Partial deletion (Exon 4 - 7)
HAMDAN ET AL - Patient 2	dn	c.1573C>T ; p.(R525*)	Nonsense mutation
CARR ET AL	dn	arr3p(70467233-71535980)x1 dn	Whole gene
O'ROAK ET AL	dn	c.1015_1016insT ; p.(A339Sfs*4)	Frameshift
TALKOWSKI ET AL	dn	46,XY,t(3;10)(p13;q21.2)	Balanced translocation - breakpoint in FOXP1
LE FEVRE ET AL	dn	arr3p13(71041636-71229421)x1 dn	Partial deletion (Exon 7 - 13)
PALUMBO ET AL	dn	arr3p(70429306-71437677)x1 dn	Partial deletion (Exon 4 - 21)
LOZANO ET AL	dn	c.1267_1268delGT ; p.(V423Hfs*37)	Frameshift
SONG ET AL	dn	c.1A>G ; p.(M1V)	Missense mutation
TUTULAN-CUNITA ET AL	NA	arr3p(65342824-77820255)x1	Whole deletion
SOLLIS ET AL - Patient 1	dn	c.1393A>G ; p.(R465G)	Missense mutation
SOLLIS ET AL - Patient 2	dn	c.1540C>T ; p.(R514C)	Missense mutation
SOLLIS ET AL - Patient 3	dn	c.1317C>G ; p.(Y439*)	Nonsense mutation
THEVENON ET AL - Patient 1 (Decipher 257047)	dn	arr3p13(71006071-73344433)x1 dn	Whole deletion
THEVENON ET AL - Patient 2 (Decipher 2059)	dn	arr3p13p12.2(71540307-82716711)x1 dn	Partial deletion (Exon 1 - 3)
THEVENON ET AL - Patient 3 (Decipher 4438)	dn	arr3p14.1p13(68541944-72842054)x1 dn	Whole deletion
DIMITROV ET AL - Patient 1	dn	arr3p(58598577-74508971)x1 dn	Whole deletion
DIMITROV ET AL - Patient 2	NA	arr3p(59703992-86824958)x1	Whole deletion
DIMITROV ET AL - Patient 3	NA	arr3p(62046934-77330720)x1	Whole deletion

Sequences were compared to the wild-type sequence as submitted to NM_032682.5 (Ensembl Accession Number ENST00000318789.4).
CNVs were mapped against the human genome build hg19/GRCh37.