

PATIENTS	REFERENCE	FAMILY	ORIGIN	CONSAQUINITY	AGE AT DIAGNOSIS (years)	Mutation 1	Mutation 2	PRENATAL FEATURES	SEX	OC AT BIRTH (SD)	WEIGHT AT BIRTH (SD)	HEIGHT AT BIRTH (SD)	AGE AT TIME OF EXAMINATION (years)	OC AT TIME OF EXAMINATION (DS)	WEIGHT AT TIME OF EXAMINATION (DS)	HEIGHT AT TIME OF EXAMINATION (DS)	BRAIN IMAGING	NEURODEVELOPMENTAL OUTCOME	EPILEPSY	OTHER SYMPTOMS	
Patient #1	Moyinhan, 2000/ Bond, 2005	#1	Pakistan	Yes	Not Provided	c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	M	Not Provided	Not Provided	Not Provided	Not Provided	-6	Not Provided	Not Provided	Not performed	Mild intellectual disability	No	No	
Patient #2	Moyinhan, 2000/ Bond, 2005	#1	Pakistan	Yes	Not Provided	c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	-8	0,0	0,0	Not performed	Moderate intellectual disability	Yes - rare tonic/clonic seizures	Profound congenital neurosensory deafness	
Patient #3	Moyinhan, 2000/ Bond, 2005	#1	Pakistan	Yes	Not Provided	c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not performed	IQ 86	No	No	
Patient #4	Henei, 1992/Moyinhan, 2000/ Bond, 2005	#1	Pakistan	Yes	Not Provided	c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not performed	IQ 89	No	Acute Lymphoblastic Leukemia causing death - skin areas of hypopigmentation	
Patient #5	Bond, 2005	#2	Pakistan	Yes	Not Provided	c.4005-15A>G (p.E386fs*4)	c.4005-15A>G (p.E386fs*4)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	-5	Not Provided	Not Provided	Not Provided	Moderate intellectual disability	Not Provided	Not Provided	
Patient #6	Bond, 2005	#2	Pakistan	Yes	Not Provided	c.4005-15A>G (p.E386fs*4)	c.4005-15A>G (p.E386fs*4)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	-7	Not Provided	Not Provided	Not Provided	Moderate intellectual disability	Not Provided	Not Provided	
Patient #7	Hassan, 2007	#3	Pakistan	Yes	18-30	c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	-4	Not Provided	Not Provided	Not Provided	IQ 51	No	No	
Patient #8	Hassan, 2007	#3	Pakistan	Yes		c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	F	Not Provided	Not Provided	Not Provided	Not Provided	-7	Not Provided	Not Provided	Not Provided	IQ 65	No	No	
Patient #9	Hassan, 2007	#3	Pakistan	Yes		c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	M	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	No	No	No	
Patient #10	Hassan, 2007	#3	Pakistan	Yes		c.246T>A (p.Ser82*)	c.246T>A (p.Ser82*)	Not Provided	M	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	No	No	No
Patient #11	Pagnamenta, 2012	#4	Somalia	Yes	6	c.700G>T (p.Glu234*)	c.700G>T (p.Glu234*)	Not Provided	F	-3,7	-2,4	-3,7	6	-8,9	-1,1	-1,6	No abnormalities	Mild intellectual disability	No	severe gastric reflux - Moderate to severe bilateral neurosensory deafness - Slight hyperlaxity - Hypotonia	
Patient #12	Issa, 2013	#5	Italy	Yes	Not Provided	c.4441C>T (p.Arg1481*)	c.4441C>T (p.Arg1481*)	Not Provided	M	Not Provided	0,5	Not Provided	5	-6	Not Provided	-4,1	Simplified gyral pattern (mostly frontal) - corpus callosum agenesis	IQ 69	No	Abdominal hernia - Skin areas of hyperpigmentation - Behavioral disorder - Tic disorder	
Patient #13	Issa, 2013	#5	Italy	Yes	Not Provided	c.4441C>T (p.Arg1481*)	c.4441C>T (p.Arg1481*)	Not Provided	M	-3,5	-0,5	-0,5	2	-5	Not Provided	Not Provided	Simplified gyral pattern (mostly frontal) - corpus callosum and cerebellar hypoplasia	IQ 56	No	Skin areas of hyperpigmentation - Behavioral disorders	
Patient #14	Lancaster, 2013	#6	Not Provided	No	Not Provided	c.4546G>T (p.Glu1516*)	c.4672C>T (p.Arg1558*)	Microcephaly	F	-6,5	-3,9	-5	3	-13,2	Not Provided	-6,7	Simplified gyral pattern	Not Provided	No	Neurosensory deafness	
Patient #15	Tan, 2014	#7	Caucasian	No	5	c.524_528del(p.Gln175Argfs*42)	c.4005-1G>A (fs*)	Microcephaly	F	Not Provided	Not Provided	Not Provided	6	-8,9	-1,5	-3	No abnormalities	IQ 75	No	Asthma - Allergies - Sleep apnea	
Patient #16	Li, 2015	#8	Hispanic	No	0,5	c.4441C>T (p.Arg1481*)	c.5227C>T (p.Gln1743*)	No	M	-3,1	-0,3	-1,5	Not Provided	Not Provided	Not Provided	Not Provided	Short corpus callosum	Not Provided	No	Failure to thrive (enteral nutrition)	
Patient #17	Yigit, 2015	#9	Turkey	Yes	Not Provided	c.4005-9A>G (p.Arg1335Serfs*3)	c.4005-9A>G (p.Arg1335Serfs*3)	No	M	Not Provided	-2	Not Provided	9	-4	Not Provided	-3,2	Not Provided	Not Provided	No	Bone age delay -5th finger clinodactyly - 11 pairs of ribs	
Patient #18	Yigit, 2015	#9	Turkey	Yes	Not Provided	c.4005-9A>G (p.Arg1335Serfs*3)	c.4005-9A>G (p.Arg1335Serfs*3)	Not Provided	F	Not Provided	Not Provided	Not Provided	10	-4	Not Provided	-3,4	No abnormalities	Mild intellectual disability	No	Congenital hip dislocation	
Patient #19	Yigit, 2015	#10	Pakistan	Yes	Not Provided	c.383+1G>C(p.Lys129*)	c.383+1G>C(p.Lys129*)	Not Provided	F	Not Provided	-2	Not Provided	2	-6,2	Not Provided	-5	No abnormalities	Not Provided	No	Not Provided	
Patient #20	Jouan, 2016	#11	Canada	No	53	c.280G>A (p.Gly94Arg)	c.3695A>G (p.Asn1232Ser)	No	F	Not Provided	Not Provided	Not Provided	53	1,0	Not Provided	0,0	Corpus callosum agenesis	IQ 84	No	Learning disabilities	
Patient #21	Jouan, 2016	#11	Canada	No	52	c.280G>A (p.Gly94Arg)	c.3695A>G (p.Asn1232Ser)	No	F	Not Provided	Not Provided	Not Provided	52	-1,3	Not Provided	-0,5	Corpus callosum agenesis	IQ 78	No	Mutism - Intermittent ataxia	
Patient #22	Jouan, 2016	#11	Canada	No	44	c.280G>A (p.Gly94Arg)	c.3695A>G (p.Asn1232Ser)	Not Provided	M	Not Provided	Not Provided	Not Provided	44	-1,3	Not Provided	0,5	Corpus callosum agenesis	IQ 77	No	Resolutive obsessive-compulsive disorder	
Patient #23	Pagnamenta, 2016	#12	Not Provided	No	11	c.4604-1G>C	c.3097delG (p.V1033 fs*41)	Not Provided	M	Not Provided	Not Provided	Not Provided	11	-5,5	Not Provided	Not Provided	No abnormalities	developmental quotient: 45	No	Café-au-lait spots	
Patient #24	Abdullah, 2017	#13	Pakistan	Yes	30	c.5127_5128dupGT (p.Ser1710CysfsTer22)	c.5127_5128dupGT (p.Ser1710CysfsTer22)	Not Provided	F	Not Provided	Not Provided	Not Provided	30	Not Provided	Not Provided	Not Provided	Not Provided	Severe intellectual disability - walking	No	No	
Patient #25	Abdullah, 2017	#13	Pakistan	Yes	32	c.5127_5128dupGT (p.Ser1710CysfsTer22)	c.5127_5128dupGT (p.Ser1710CysfsTer22)	Not Provided	M	Not Provided	Not Provided	Not Provided	32	Not Provided	Not Provided	Not Provided	Not Provided	Severe intellectual disability - walking	No	Moderate hearing impairment	
Patient #26	Abdullah, 2017	#13	Pakistan	Yes	12	c.5127_5128dupGT (p.Ser1710CysfsTer22)	c.5127_5128dupGT (p.Ser1710CysfsTer22)	Not Provided	M	Not Provided	Not Provided	Not Provided	12	Not Provided	Not Provided	Not Provided	Not Provided	Moderate intellectual disability	No	No	
Patient #27	Abdullah, 2017	#13	Pakistan	Yes	8	c.5127_5128dupGT (p.Ser1710CysfsTer22)	c.5127_5128dupGT (p.Ser1710CysfsTer22)	Not Provided	F	Not Provided	Not Provided	Not Provided	8	Not Provided	Not Provided	Not Provided	Not Provided	Moderate intellectual disability	No	No	
Patient #28	Ahmad, 2017	#14	Pakistan	Yes	Not Provided	c.1279C>T (p.Arg427*)	c.1279C>T (p.Arg427*)	Not Provided	NP	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided
Patient #29	Sukumaran, 2017	#15	Pakistan	Yes	16	c.4114C>T (p.Arg1372*)	c.4114C>T (p.Arg1372*)	Not Provided	F	Not Provided	Not Provided	Not Provided	16	-5	Not Provided	Not Provided	Not Provided	Mild intellectual disability	No	No	
Patient #30	Sukumaran, 2017	#15	Pakistan	Yes	12	c.4114C>T (p.Arg1372*)	c.4114C>T (p.Arg1372*)	Not Provided	M	Not Provided	Not Provided	Not Provided	12	-9	Not Provided	Not Provided	Not Provided	Mild intellectual disability	No	No	
Patient #31	Alfares, 2018	#16	Saudi Arabia	Yes	16	c.4055A>G (p.Glu1352Gln)	c.4055A>G (p.Glu1352Gln)	Not Provided	M	-2	1100	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	White matter abnormalities	Severe intellectual disability - non walking	Yes - epileptic encephalopathy with hypersarhythmia	Bilateral cataract - White matter abnormalities - Tetrapyramidal syndrome
Patient #32	Alfares, 2018	#16	Saudi Arabia	Yes	10	c.4055A>G (p.Glu1352Gln)	c.4055A>G (p.Glu1352Gln)	Not Provided	M	Not Provided	Not Provided	Not Provided	10	-2	-4	-4	moderate ventriculomegaly	Severe intellectual disability - non walking	Yes	Bilateral cataract - Tetrapyramidal syndrome - Dystonia	
Patient #33	Alfares, 2018	#16	Saudi Arabia	Yes	8	c.4055A>G (p.Glu1352Gln)	c.4055A>G (p.Glu1352Gln)	Not Provided	F	Not Provided	Not Provided	Not Provided	8	-4	-4,5	-5	Thin corpus callosum	Severe intellectual disability - non walking	Yes - epileptic encephalopathy with hypersarhythmia	Bilateral cataract - Tetrapyramidal syndrome	
Patient #34	Alfares, 2018	#16	Saudi Arabia	Yes	6	c.4055A>G (p.Glu1352Gln)	c.4055A>G (p.Glu1352Gln)	Intra uterine growth retardation	M	-2	1700	Not Provided	0,75	-4	Not Provided	Not Provided	Thin corpus callosum	Not Provided	Yes	Bilateral cataract - Hypospadias - Bilateral cryptorchidism - Cardiac congenital defect	
Patient #35	Alfares, 2018	#17	Saudi Arabia	Not Provided	3	c.4672C>T (p.Arg1558*)	c.4672C>T (p.Arg1558*)	Not Provided	M	-3,5	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Not Provided	Decreased cerebral volume	Intellectual disability	Yes	Cataract	