

Table S2 Detailed Clinical Features

	<i>Male (M) / Female (F)</i>	<i>Term at birth (weeks)</i>	<i>Birth weight</i>	<i>Birth length</i>	<i>Birth head circumference</i>
Patient 1	F	At term	2740 g (-1.3 SD)(3-10th Centile)	47.5 cm (-0.7 SD)(3-10th Centile)	33.5 cm (-0.7 SD)(10th Centile)
Patient 2	M	36	3230 g (+2 SD)(90th Centile)	48.5 cm (+0.5 SD)(50-90th Centile)	35 cm (+2 SD)(90-97th Centile)
Patient 3	M	34	2180 g (0 SD) (10-50th Centile)	nd	nd
Patient 4	M	At term	3100 g (-1 SD)(10-50th Centile)	49.5 cm (-0.5 SD)(10-50th Centile)	38 cm (+2 SD)(90-97th Centile)
Patient 5	M	At term	3160 g (10-50th Centile)	51 cm (50th Centile)	nd
Patient 6	M	At term	3487 g (50th Centile)	nd	nd
Patient 7 (Decipher 331530)	M	37	3670 g (>90th Centile)	53 cm (>97th Centile)	36.5 cm (97th Centile)
Patient 8	M	31	1885 g (50-90th Centile)	39.5 cm (10-50th Centile)	nd
Patient 9 (Decipher 255792)	M	28+1	1150 g (25-50th Centile)	nd	27 cm (50-75th Centile)
Patient 10 (Decipher 260107)	M	At term	2770 g (-1.94 SD)	51 cm (-0.65 SD)	33 cm (-2 SD)
Patient 11 (Decipher 272645)	M	At term	3544 g	50.8 cm	nd
Patient 12 (Decipher 274063)	M	36+1	2500 g (3th Centile)	46 cm (3th Centile)	35 cm (50th Centile)
Patient 13 (Decipher 284583)	F	At term	3330 g	50 cm	nd
Patient 14 (Decipher 272245)	M	34	2220 g	nd	nd
Patient 15 (Decipher 274751)	F	At term	nd	nd	nd
Patient 16 (Decipher 262739)	M	At term	50-75th Centile	nd	nd
Patient 17 (Decipher 279456)	F	At term	25-50th Centile	nd	98th Centile
Patient 18 (Decipher 270887)	M	At term	3010 g	nd	nd
Patient 19 (Decipher 252324)	F	At term	3400 g (10-50th Centile)	nd	nd
Patient 20	M	At term	2980 g	nd	nd
Patient 21	M	At term	3310 g	49 cm	36 cm
Patient 22	M	At term	3680 g	51 cm	36 cm
Patient 23	M	At term	3040 g	47 cm	33 cm
Patient 24	M	32	1160 g	nd	nd
Patient 25	M	At term	3520 g (50th Centile)	52 cm (75th Centile)	34 cm (50th Centile)

	Male (M) / Female (F)	Term at birth (weeks)	Birth weight	Birth length	Birth head circumference
PARIANI ET AL	M	At term	3123 g (-1 SD)	nd	nd
HORN ET AL - Patient 1	M	nd	nd	nd	nd
HORN ET AL - Patient 2	F	nd	nd	nd	nd
HORN ET AL - Patient 3	M	nd	nd	nd	nd
HAMDAN ET AL - Patient 1	F	nd	nd	nd	nd
HAMDAN ET AL - Patient 2	M	nd	nd	nd	nd
CARR ET AL	M	At term	N	N	N
O'ROAK ET AL	M	At term	nd	nd	nd
TALKOWSKI ET AL	M	nd	nd	nd	nd
LE FEVRE ET AL	M	At term	3200 g (25th Centile)	48 cm (25th Centile)	36.5 cm (75th Centile)
PALUMBO ET AL	M	At term	3220 g (25th Centile)	50 cm (25th Centile)	35 cm (50th Centile)
LOZANO ET AL	F	nd	nd	nd	nd
SONG ET AL	F	34	2100 g (+0.1 SD)	nd	nd
TUTULAN-CUNITA ET AL *	M	At term	3500 g	nd	nd
SOLLIS ET AL - Patient 1	M	nd	nd	nd	nd
SOLLIS ET AL - Patient 2	M	nd	nd	nd	nd
SOLLIS ET AL - Patient 3	F	At term	4160 g	nd	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	F	34+5	2250 g (5th Centile)	44 cm (3th Centile)	32.5 cm (25-50th Centile)
THEVENON ET AL - Patient 2 (Decipher 2059)	F	nd	nd	nd	nd
THEVENON ET AL - Patient 3 (Decipher 4438)	M	At term	2680 g (<5th Centile)	49 cm (10-25th Centile)	34 cm (25-50th Centile)
DIMITROV ET AL - Patient 1	F	At term	2040 g (0.4-2nd Centile)	nd	nd
DIMITROV ET AL - Patient 2	M	At term	2320 g (-1.2 SD)	46 cm (-1.6 SD)	32 cm (-2.6 SD)
DIMITROV ET AL - Patient 3	F	At term	2200 g (0.4-2nd Centile)	nd	nd

* additional data from referring clinician

	<i>Age at last follow-up (Years)</i>	<i>Weight (Centile)</i>	<i>Length (Centile)</i>	<i>BMI (Centile)</i>	<i>Head circumference (Centile)</i>	<i>Mild / moderate NMD</i>	<i>Severe NMD</i>	<i>Mild / moderate ID</i>	<i>Severe ID</i>
Patient 1	2.5	< 3	3-10	10	25-50	+	-	+	-
Patient 2	2.5	25	10-25	75	90	+	-	+	-
Patient 3	16	< 3	< 3	< 3	> 97	-	+	-	+
Patient 4	1.67	25	10	50	50	+	-	+	-
Patient 5	3	< 3	25	< 3	10-25	+	-	+	-
Patient 6	14	3-10	< 3	25-50	50	+	-	+	-
Patient 7 (Decipher 331530)	3.5	25-50	50	25	97	+	-	+	-
Patient 8	15	50	25	75-90	> 97	-	+	-	+
Patient 9 (Decipher 255792)	0.42	nd	nd	nd	nd	nd	nd	nd	nd
Patient 10 (Decipher 260107)	4	< 3	< 3	25	50	-	+	-	+
Patient 11 (Decipher 272645)	5	10-25	3-10	50	nd	+	-	+	-
Patient 12 (Decipher 274063)	7.5	< 3	< 3	25-50	nd	-	+	-	+
Patient 13 (Decipher 284583)	5	25-50	20	50	25	+	-	+	-
Patient 14 (Decipher 272245)	9.5	10-25	50	nd	10	+	-	+	-
Patient 15 (Decipher 274751)	23	nd	nd	nd	> 97	+	-	+	-
Patient 16 (Decipher 262739)	17	50-75	75	nd	90	+	-	-	+
Patient 17 (Decipher 279456)	5	50-75	50	nd	> 97	-	-	+	-
Patient 18 (Decipher 270887)	10	< 3	< 3	10	< 3	-	+	-	+
Patient 19 (Decipher 252324)	2	3	10	3-10	25-50	+	-	+	-
Patient 20	5	25-50	3-10	75-90	nd	+	-	+	-
Patient 21	12	< 3	< 3	10-25	97	+	-	+	-
Patient 22	9	50	50-75	25-50	97	-	-	-	-
Patient 23	10	3	10	10-25	25	-	+	-	+
Patient 24	7	< 3	< 3	3-10	< 3	-	+	-	+
Patient 25	8.5	50-75	50-75	50	50	+	-	+	-

	<i>Age at last follow-up (Years)</i>	<i>Weight (Centile)</i>	<i>Length (Centile)</i>	<i>BMI (Centile)</i>	<i>Head circumference (Centile)</i>	<i>Mild / moderate NMD</i>	<i>Severe NMD</i>	<i>Mild / moderate ID</i>	<i>Severe ID</i>
PARIANI ET AL	1.9	< 10	< 2	nd	50	+	-	nd	nd
HORN ET AL - Patient 1	7	40	80	nd	75	+	-	+	-
HORN ET AL - Patient 2	5.5	> 98	45	nd	60	+	-	+	-
HORN ET AL - Patient 3	6	> 98	30	nd	80	+	-	+	-
HAMDAN ET AL - Patient 1	15	nd	nd	nd	nd	+	-	+	-
HAMDAN ET AL - Patient 2	9	nd	nd	nd	nd	+	-	+	-
CARR ET AL	3.4	10-25	10	nd	50-75	+	-	nd	nd
O'ROAK ET AL	nd	nd	nd	nd	nd	nd	nd	+	-
TALKOWSKI ET AL	nd	nd	nd	nd	nd	+	-	+	-
LE FEVRE ET AL	7	40	25	nd	> 98	+	-	+	-
PALUMBO ET AL	20	N	N	nd	N	+	-	-	+
LOZANO ET AL	14	75	58	nd	> 98	+	-	+	-
SONG ET AL	22	25	< 2	nd	nd	+	-	+	-
TUTULAN-CUNITA ET AL *	5	50	15	nd	15-50	-	+	-	+
SOLLIS ET AL - Patient 1	11	nd	↓	nd	↑	+	-	+	-
SOLLIS ET AL - Patient 2	7	N	N	nd	N	+	-	+	-
SOLLIS ET AL - Patient 3	15	nd	N	nd	N	+	-	-	+
THEVENON ET AL - Patient 1 (Decipher 257047)	3	nd	nd	nd	nd	-	+	-	+
THEVENON ET AL - Patient 2 (Decipher 2059)	19	nd	nd	nd	nd	nd	nd	+	-
THEVENON ET AL - Patient 3 (Decipher 4438)	3	< 5	nd	nd	nd	-	+	-	+
DIMITROV ET AL - Patient 1	4	25-50	< 0.4	nd	50	-	+	-	+
DIMITROV ET AL - Patient 2	0.3	< 3	< 0.4	nd	< 3	-	+	nd	nd
DIMITROV ET AL - Patient 3	1	< 0.4	< 0.4	nd	< 0.4	-	+	nd	nd

	<i>Speech and language delay</i>	<i>Expressive > receptive</i>	<i>First words (Years)</i>	<i>Combined words (Years)</i>	<i>Articulation problems</i>	<i>Poor grammar</i>	<i>OMD</i>	<i>Behavioral difficulties</i>	<i>Autistic features</i>
Patient 1	+	+	> 3	nd	+	+	+	-	+
Patient 2	+	+	Nil	/	/	/	nd	-	-
Patient 3	+	+	Nil	/	/	/	nd	+	nd
Patient 4	+	+	Nil	/	/	/	-	+	-
Patient 5	+	+	Nil	/	/	/	+	-	+
Patient 6	+	nd	3	+ age?	+	nd	nd	+	nd
Patient 7 (Decipher 331530)	+	+	3	nd	nd	nd	nd	+	+
Patient 8	+	+	1	1.3	+	+	+	+	+
Patient 9 (Decipher 255792)	nd	nd	nd	nd	nd	nd	nd	nd	nd
Patient 10 (Decipher 260107)	+	+	Nil	/	nd	nd	+	nd	+
Patient 11 (Decipher 272645)	+	+	2	3	+	nd	nd	-	nd
Patient 12 (Decipher 274063)	+	+	4	nd	+	+	+	nd	-
Patient 13 (Decipher 284583)	+	+	2	3	+	+	+	-	+
Patient 14 (Decipher 272245)	+	nd	1.5	nd	+	nd	-	-	-
Patient 15 (Decipher 274751)	+	nd	2.5	nd	nd	nd	-	+	+
Patient 16 (Decipher 262739)	+	nd	2	9	-	nd	-	+	+
Patient 17 (Decipher 279456)	+	+	nd	nd	nd	nd	-	+	-
Patient 18 (Decipher 270887)	+	+	Nil	/	nd	nd	nd	nd	nd
Patient 19 (Decipher 252324)	+	+	2	3	+	-	+	+	-
Patient 20	+	+	Nil	/	/	/	+	+	+
Patient 21	nd	nd	nd	nd	nd	nd	nd	+	+
Patient 22	+	-	1.8	1.8	-	-	-	-	+
Patient 23	+	+	Nil	/	/	/	-	nd	+
Patient 24	+	+	Nil	/	/	/	+	nd	+
Patient 25	+	+	+ regression	-	+	+	+	+	+

	<i>Tonicity</i>	<i>Spasticity / contractures</i>	<i>Reflexes</i>	<i>Seizures</i>	<i>Brain imaging</i>	<i>FTT</i>	<i>Feeding difficulties</i>	<i>Obesity</i>
Patient 1	↓	-	N	-	N	-	+	-
Patient 2	↓ / ↑	+	N	-	+	-	-	-
Patient 3	↓ / ↑	+	nd	+	N	+	+	-
Patient 4	↓	-	N	+	N	+	+	-
Patient 5	nd	nd	nd	nd	N	-	+	-
Patient 6	↓	nd	nd	nd	N	nd	nd	nd
Patient 7 (Decipher 331530)	nd	nd	nd	-	+	-	-	-
Patient 8	↑	+	↓	-	N	+	+	-
Patient 9 (Decipher 255792)	nd	nd	nd	nd	+	nd	+	nd
Patient 10 (Decipher 260107)	↓	+	N	-	N	+	+	-
Patient 11 (Decipher 272645)	↑	+	N	-	N	-	+	-
Patient 12 (Decipher 274063)	↓	-	N	-	nd	+	+	-
Patient 13 (Decipher 284583)	N	-	N	-	N	-	+	-
Patient 14 (Decipher 272245)	N	-	N	-	nd	+	+	-
Patient 15 (Decipher 274751)	N	-	N	-	+	-	+	-
Patient 16 (Decipher 262739)	nd	nd	nd	-	+	-	-	-
Patient 17 (Decipher 279456)	N	-	N	-	+	-	-	-
Patient 18 (Decipher 270887)	↓	+	nd	-	+	nd	+	-
Patient 19 (Decipher 252324)	N	-	N	-	+	+	+	-
Patient 20	N	+	N	-	+	-	-	-
Patient 21	↓	+	N	-	+	-	-	-
Patient 22	N	-	N	+	N	-	-	-
Patient 23	↑	+	↑	-	N	-	-	-
Patient 24	↑	+	↑	-	N	+	+	-
Patient 25	↓	nd	nd	-	+	-	+	-

	Tonicity	Spasticity / contractures	Reflexes	Seizures	Brain imaging	FTT	Feeding difficulties	Obesity
PARIANI ET AL	↑	+	N	-	+	+	+	-
HORN ET AL - Patient 1	nd	nd	nd	-	N	-	nd	-
HORN ET AL - Patient 2	nd	nd	nd	-	N	-	+	+
HORN ET AL - Patient 3	nd	nd	nd	-	N	-	-	+
HAMDAN ET AL - Patient 1	nd	nd	nd	nd	nd	nd	nd	nd
HAMDAN ET AL - Patient 2	nd	nd	nd	nd	nd	nd	nd	nd
CARR ET AL	↓ / ↑	-	↑	+	+	-	-	-
O'ROAK ET AL	nd	nd	nd	+	nd	nd	nd	nd
TALKOWSKI ET AL	nd	nd	nd	nd	nd	nd	nd	nd
LE FEVRE ET AL	N	-	N	-	+	+	+	-
PALUMBO ET AL	nd	nd	nd	nd	N	nd	nd	-
LOZANO ET AL	↓	nd	nd	nd	+	-	nd	-
SONG ET AL	N	nd	N	-	nd	-	nd	-
TUTULAN-CUNITA ET AL *	↑	+	↑	-	+	-	-	-
SOLLIS ET AL - Patient 1	↓	nd	nd	nd	+	nd	nd	nd
SOLLIS ET AL - Patient 2	↓	nd	nd	nd	nd	-	nd	-
SOLLIS ET AL - Patient 3	↓	nd	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	↓	+	nd	+	+	+	+	nd
THEVENON ET AL - Patient 2 (Decipher 2059)	↓	+	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 3 (Decipher 4438)	nd	+	nd	nd	nd	+	+	nd
DIMITROV ET AL - Patient 1	↑	+	↑	nd	+	+	+	nd
DIMITROV ET AL - Patient 2	↑	+	↑	+	+	+	+	nd
DIMITROV ET AL - Patient 3	↑	+	↑	nd	+	+	+	nd

	<i>Broad, prominent forehead</i>	<i>Frontal hair upsweep</i>	<i>Hypertelorism</i>	<i>Downslanting PF</i>	<i>Bent PF</i>	<i>Ptosis</i>	<i>Blepharophimosis</i>	<i>Short nose</i>	<i>Broad nose tip</i>	<i>Pronounced nasolabial folds</i>
Patient 1	+	+	-	+	+	+	-	+	+	+
Patient 2	+	+	+	+	+	+	-	+	+	+
Patient 3	+	+	-	+	-	-	+	+	+	-
Patient 4	+	-	-	-	-	+	-	+	+	+
Patient 5	+	-	-	-	-	+	-	+	+	-
Patient 6	-	-	+	+	-	+	-	-	+	-
Patient 7 (Decipher 331530)	+	-	-	+	-	+	-	-	+	+
Patient 8	+	+	+	-	-	+	-	+	+	+
Patient 9 (Decipher 255792)	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
Patient 10 (Decipher 260107)	+	-	+	+	-	-	-	+	+	-
Patient 11 (Decipher 272645)	+	-	+	-	+	+	-	+	+	+
Patient 12 (Decipher 274063)	+	+	+	+		+	+	+	+	-
Patient 13 (Decipher 284583)	+	+	-	+		-	-	+	+	+
Patient 14 (Decipher 272245)	+	-	-	+		+	-	+	+	-
Patient 15 (Decipher 274751)	+	-	-	-		-	-	-	-	-
Patient 16 (Decipher 262739)	+	-	-	+		-	-	-	-	-
Patient 17 (Decipher 279456)	+	-	+	-		+	-	+	+	-
Patient 18 (Decipher 270887)	+	+	-	+	-	+	+	+	-	-
Patient 19 (Decipher 252324)	+	-	-	+	-	+	+	+	+	-
Patient 20	+	+	+	-	+	+	-	+	-	+
Patient 21	+	-	-	-	-	-	-	-	+	+
Patient 22	+	-	+	-	-	-	-	+	-	-
Patient 23	+	+	-	+		-	-	-	+	-
Patient 24	+	+	+	-		+	-	-	+	+
Patient 25	+	-	+	+	+	+	-	+	+	-

	<i>Broad, prominent forehead</i>	<i>Frontal hair upsweep</i>	<i>Hypertelorism</i>	<i>Downslanting PF</i>	<i>Bent PF</i>	<i>Ptosis</i>	<i>Blepharophimosis</i>	<i>Short nose</i>	<i>Broad nose tip</i>	<i>Pronounced nasolabial folds</i>
PARIANI ET AL	-	-	-	-	nd	-	+	+	+	-
HORN ET AL - Patient 1	-	-	-	-	nd	-	-	-	-	-
HORN ET AL - Patient 2	+	+	-	-	nd	+	-	+	+	-
HORN ET AL - Patient 3	+	+	-	+	nd	+	-	+	+	-
HAMDAN ET AL - Patient 1	-	-	nd	-	nd	nd	nd	-	-	nd
HAMDAN ET AL - Patient 2	-	-	nd	-	nd	nd	nd	-	-	nd
CARR ET AL	+	-	+	+	nd	+	-	+	+	-
O'ROAK ET AL	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
TALKOWSKI ET AL	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
LE FEVRE ET AL	+	-	-	+	nd	-	-	+	+	-
PALUMBO ET AL	+	-	-	+	nd	+	-	-	-	-
LOZANO ET AL	nd	+	-	+	nd	+	-	-	+	+
SONG ET AL	+	+	nd	-	nd	nd	nd	+	+	nd
TUTULAN-CUNITA ET AL *	+	-	+	-	nd	+	-	+	+	-
SOLLIS ET AL - Patient 1	+	nd	nd	nd	nd	nd	nd	nd	nd	nd
SOLLIS ET AL - Patient 2	nd	nd	+	+	nd	nd	nd	+	nd	nd
SOLLIS ET AL - Patient 3	+	nd	+	nd	nd	nd	nd	nd	+	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	nd	nd	+	nd	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 2 (Decipher 2059)	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 3 (Decipher 4438)	nd	nd	+	nd	nd	nd	-	nd	nd	nd
DIMITROV ET AL - Patient 1	+	+	+	+	nd	nd	nd	+	nd	nd
DIMITROV ET AL - Patient 2	+	-	+	nd	nd	nd	-	+	-	nd
DIMITROV ET AL - Patient 3	nd	nd	+	nd	nd	nd	nd	nd	-	nd

	<i>Wide mouth with full lips</i>	<i>Pronounced vermilion border</i>	<i>Downturned corners of the mouth</i>	<i>Open mouth</i>	<i>Prominent chin</i>	<i>Chin with horizontal crease</i>	<i>Hypoplastic upper ear helix</i>	<i>Prominent finger pads</i>	<i>Single palmar crease</i>	<i>Clinodactyly</i>
Patient 1	+	+	-	nd	-	+	-	-	+	-
Patient 2	+	+	-	nd	-	+	+	-	-	-
Patient 3	-	+	+	nd	+	-	-	-	-	-
Patient 4	+	+	-	-	+	+	-	-	-	+
Patient 5	-	+	+	+	-	+	-	-	+	-
Patient 6	+	+	+	+	+	+	+	+	nd	nd
Patient 7 (Decipher 331530)	+	+	+	+	-	+	-	-	-	-
Patient 8	+	+	+	+	+	+	+	-	+	+
Patient 9 (Decipher 255792)	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
Patient 10 (Decipher 260107)	-	+	+	-	-	+	+	-	+	+
Patient 11 (Decipher 272645)	+	-	-	nd	+	+	-	nd	+	nd
Patient 12 (Decipher 274063)	-	nd	-	-	nd	nd	-	-	+	-
Patient 13 (Decipher 284583)	-	-	+	+	-	-	-	-	-	-
Patient 14 (Decipher 272245)	+	nd	-	+	nd	nd	-	-	-	+
Patient 15 (Decipher 274751)	-	-	-	-	-	-	-	-	-	-
Patient 16 (Decipher 262739)	-	-	-	-	-	-	-	+	+	-
Patient 17 (Decipher 279456)	+	+	+	+	-	-	-	-	+	-
Patient 18 (Decipher 270887)	-	+	+	-	+	-	-	-	nd	-
Patient 19 (Decipher 252324)	-	+	+	+	-	-	+	-	-	+
Patient 20	+	+	+	+	-	-	-	-	-	-
Patient 21	+	+	-	+	+	-	-	nd	nd	nd
Patient 22	+	-	-	-	-	-	-	-	-	-
Patient 23	+	-	+	-	-	-	-	-	-	-
Patient 24	-	+	+	+	+	+	-	-	-	+
Patient 25	+	+	+	+	+	-	-	-	-	-

	<i>Wide mouth with full lips</i>	<i>Pronounced vermillion border</i>	<i>Downturned corners of the mouth</i>	<i>Open mouth</i>	<i>Prominent chin</i>	<i>Chin with horizontal crease</i>	<i>Hypoplastic upper ear helix</i>	<i>Prominent finger pads</i>	<i>Single palmar crease</i>	<i>Clinodactyly</i>
PARIANI ET AL	-	nd	+	+	nd	nd	nd	nd	+	+
HORN ET AL - Patient 1	-	nd	-	+	nd	nd	-	-	-	-
HORN ET AL - Patient 2	-	nd	-	+	nd	nd	-	-	-	-
HORN ET AL - Patient 3	-	nd	-	+	nd	nd	-	-	-	-
HAMDAN ET AL - Patient 1	nd	nd	nd	nd	nd	nd	nd	-	-	-
HAMDAN ET AL - Patient 2	nd	nd	nd	nd	nd	nd	nd	-	-	-
CARR ET AL	-	nd	+	-	nd	nd	-	+	+	-
O'ROAK ET AL	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
TALKOWSKI ET AL	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
LE FEVRE ET AL	-	nd	-	-	nd	nd	-	+	-	+
PALUMBO ET AL	+	nd	-	+	nd	nd	-	-	-	-
LOZANO ET AL	+	nd	+	nd	nd	nd	-	nd	nd	nd
SONG ET AL	nd	nd	nd	nd	nd	nd	nd	-	-	-
TUTULAN-CUNITA ET AL *	-	nd	-	+	nd	nd	+	-	-	-
SOLLIS ET AL - Patient 1	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
SOLLIS ET AL - Patient 2	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
SOLLIS ET AL - Patient 3	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	-	nd	nd	nd	nd	nd	-	nd	-	nd
THEVENON ET AL - Patient 2 (Decipher 2059)	nd	nd	nd	nd	nd	nd	nd	nd	nd	nd
THEVENON ET AL - Patient 3 (Decipher 4438)	-	nd	nd	nd	nd	nd	-	nd	nd	nd
DIMITROV ET AL - Patient 1	-	nd	+	nd	nd	nd	+	nd	+	-
DIMITROV ET AL - Patient 2	-	nd	nd	-	nd	nd	+	nd	+	-
DIMITROV ET AL - Patient 3	-	nd	nd	nd	nd	nd	-	nd	+	-

Other congenital malformations

Patient 1	-
Patient 2	Facial hemangioma.
Patient 3	-
Patient 4	Congenital hip dislocation. Mild syndactyly II-III.
Patient 5	-
Patient 6	Hemangioma on back. Mild scoliosis. Hindfoot valgus deformity.
Patient 7 (Decipher 331530)	Facial asymmetry.
Patient 8	Bilateral inguinal herniation. Abnormal larynx. Talipes cavus. Nasolacrimal duct abnormality. Dolicocephaly. Cerebral palsy.
Patient 9 (Decipher 255792)	Diaphragmatic hernia
Patient 10 (Decipher 260107)	Bilateral severe hip dysplasia. Bilateral pes planus.
Patient 11 (Decipher 272645)	Bilateral inguinal hernia. 2/3 toe syndactyly bilateral.
Patient 12 (Decipher 274063)	Fronto-nasal angioma. Wide-spaced nipples
Patient 13 (Decipher 284583)	Highly arched palate. Single upper incisor. Plagiocephaly.
Patient 14 (Decipher 272245)	Sacral dimple. Poorly developed scrotal sac. Neonatal conjugated hyperbilirubinemia.
Patient 15 (Decipher 274751)	Spaced frontal incisors. Velopharyngeal incompetence.
Patient 16 (Decipher 262739)	Duane anomaly. Anal fistula. Ear pit.
Patient 17 (Decipher 279456)	Spina bifida occulta.
Patient 18 (Decipher 270887)	Inguinal hernia. Congenital absence of foreskin.
Patient 19 (Decipher 252324)	-
Patient 20	-
Patient 21	Unilateral ear pits.
Patient 22	-
Patient 23	-
Patient 24	Club foot. Inguinal hernia.
Patient 25	Trigonocephaly. Facial asymmetry. Sacral dimple.

PARIANI ET AL	Contractures of hand and feet.
HORN ET AL - Patient 1	-
HORN ET AL - Patient 2	-
HORN ET AL - Patient 3	-
HAMDAN ET AL - Patient 1	-
HAMDAN ET AL - Patient 2	Atresia of the midjejunum and midileum gut.
CARR ET AL	Chiari 1 malformation.
O'ROAK ET AL	-
TALKOWSKI ET AL	Spina bifida occulta. Bilateral inguinal hernia.
LE FEVRE ET AL	-
PALUMBO ET AL	Abnormal tooth position with malocclusion.
LOZANO ET AL	Highly arched palate. Wide-spaced teeth. Single umbilical artery.
SONG ET AL	-
TUTULAN-CUNITA ET AL *	Hand and feet malformations. Sacral sinus.
SOLLIS ET AL - Patient 1	-
SOLLIS ET AL - Patient 2	Mild retrognathia.
SOLLIS ET AL - Patient 3	-
THEVENON ET AL - Patient 1 (Decipher 257047)	Mild microstomy. Highly arched palate. Distal limb contractures. Absent palmar flexion creases on fingers.
THEVENON ET AL - Patient 2 (Decipher 2059)	Club feet. Bilateral hip dislocation.
THEVENON ET AL - Patient 3 (Decipher 4438)	Narrow auditory meatus. Microstomy. Congenital contractures of fingers and metatarsus varus.
DIMITROV ET AL - Patient 1	Small mouth. Chest deformaties. Hypoplastic nipples. Sacral dimple. Small hand and feet. Camptodactyly. Wide sandal gap. Club foot.
DIMITROV ET AL - Patient 2	Micrognathia. High and vaulted palate. Camptodactyly. CHARGE-like. Pes planus congenitas, bilateral sandal gaps, and sacral dimple.
DIMITROV ET AL - Patient 3	CHARGE-like: Bilateral choanal atresia, bilateral underriding 3rd toe, hypoplastic nipples, slightly anteriorly placed anus.

Other congenital malformations

	<i>Ophthalmologic abnormalities</i>	<i>Auditory abnormalities</i>
Patient 1	Strabismus, hypermetropia	-
Patient 2	nd	-
Patient 3	Nystagmus	nd
Patient 4	-	-
Patient 5	Strabismus, amblyopia	nd
Patient 6	nd	nd
Patient 7 (Decipher 331530)	Strabismus	nd
Patient 8	nd	nd
Patient 9 (Decipher 255792)	-	nd
Patient 10 (Decipher 260107)	-	Bilateral deafness (cochlear implants)
Patient 11 (Decipher 272645)	Strabismus, hypermetropia	nd
Patient 12 (Decipher 274063)	-	nd
Patient 13 (Decipher 284583)	Strabismus, right optic nerve hypoplasia	-
Patient 14 (Decipher 272245)	Strabismus	nd
Patient 15 (Decipher 274751)	Strabismus, hypermetropia	nd
Patient 16 (Decipher 262739)	Strabismus	nd
Patient 17 (Decipher 279456)	Strabismus, hypermetropia	nd
Patient 18 (Decipher 270887)	nd	+
Patient 19 (Decipher 252324)	Hypermetropia	nd
Patient 20	Strabismus, hypermetropia	-
Patient 21	Strabismus, hypermetropia	nd
Patient 22	Hypermetropia	-
Patient 23	Strabismus	-
Patient 24	Strabismus	-
Patient 25	Strabism, hypermetropia, swinging nystagmus	-

	<i>Ophthalmologic abnormalities</i>	<i>Auditory abnormalities</i>
PARIANI ET AL	Hypermetropia	nd
HORN ET AL - Patient 1	Myopia	-
HORN ET AL - Patient 2	-	-
HORN ET AL - Patient 3	-	-
HAMDAN ET AL - Patient 1	nd	nd
HAMDAN ET AL - Patient 2	nd	nd
CARR ET AL	Refractive error (picture)	-
O'ROAK ET AL	nd	nd
TALKOWSKI ET AL	nd	nd
LE FEVRE ET AL	-	-
PALUMBO ET AL	nd	nd
LOZANO ET AL	nd	nd
SONG ET AL	-	-
TUTULAN-CUNITA ET AL *	Strabismus	Bilateral deafness - sclerosis of inner ear bone chain
SOLLIS ET AL - Patient 1	nd	nd
SOLLIS ET AL - Patient 2	Strabismus	nd
SOLLIS ET AL - Patient 3	Nystagmus, hypermetropia	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	Nystagmus and bilateral strabismus	nd
THEVENON ET AL - Patient 2 (Decipher 2059)	nd	Sensorineural deafness
THEVENON ET AL - Patient 3 (Decipher 4438)	nd	nd
DIMITROV ET AL - Patient 1	Bilateral aniridia	Bilateral sensorineural hearing loss - inner ear malformation
DIMITROV ET AL - Patient 2	Bilateral microphthalmia and iris coloboma	nd
DIMITROV ET AL - Patient 3	Bilateral iris coloboma	nd

	CHD	Renal abnormalities
Patient 1	-	nd
Patient 2	Asymmetric tricuspid aortic valve with normal function	nd
Patient 3	nd	nd
Patient 4	-	-
Patient 5	nd	nd
Patient 6	nd	nd
Patient 7 (Decipher 331530)	nd	Duplex kidneys with ureterocoele and kidney dysplasia
Patient 8	nd	nd
Patient 9 (Decipher 255792)	Pulmonary hypertension, PDA	nd
Patient 10 (Decipher 260107)	Secundum ASD	-
Patient 11 (Decipher 272645)	nd	nd
Patient 12 (Decipher 274063)	-	nd
Patient 13 (Decipher 284583)	-	-
Patient 14 (Decipher 272245)	Secundum ASD	nd
Patient 15 (Decipher 274751)	ASD	nd
Patient 16 (Decipher 262739)	-	nd
Patient 17 (Decipher 279456)	ASD	nd
Patient 18 (Decipher 270887)	Secundum ASD, PS, right ventricular hypertrophy	nd
Patient 19 (Decipher 252324)	-	nd
Patient 20	Secundum ASD	nd
Patient 21	-	-
Patient 22	-	-
Patient 23	-	-
Patient 24	-	-
Patient 25	Foramen ovale apertum	-

	CHD	Renal abnormalities
PARIANI ET AL	nd	nd
HORN ET AL - Patient 1	nd	nd
HORN ET AL - Patient 2	nd	nd
HORN ET AL - Patient 3	nd	nd
HAMDAN ET AL - Patient 1	nd	nd
HAMDAN ET AL - Patient 2	nd	nd
CARR ET AL	-	-
O'ROAK ET AL	nd	nd
TALKOWSKI ET AL	nd	nd
LE FEVRE ET AL	nd	-
PALUMBO ET AL	nd	nd
LOZANO ET AL	nd	nd
SONG ET AL	nd	nd
TUTULAN-CUNITA ET AL *	ASD	N
SOLLIS ET AL - Patient 1	nd	nd
SOLLIS ET AL - Patient 2	nd	nd
SOLLIS ET AL - Patient 3	nd	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	ASD with pulmonary hypertension	nd
THEVENON ET AL - Patient 2 (Decipher 2059)	nd	nd
THEVENON ET AL - Patient 3 (Decipher 4438)	-	nd
DIMITROV ET AL - Patient 1	PDA, Secundum ASD	nd
DIMITROV ET AL - Patient 2	ASD, VSD, mild PS	nd
DIMITROV ET AL- Patient 3	PDA, ASD, pulmonary hypertension	nd

	GTA	Other organ problems
Patient 1	-	nd
Patient 2	-	nd
Patient 3	nd	nd
Patient 4	nd	Cholestasis, ductal paucity, fibrosis and chronic hepatitis
Patient 5	Bilateral cryptorchidism	nd
Patient 6	Bilateral cryptorchidism	nd
Patient 7 (Decipher 331530)	Bilateral cryptorchidism	nd
Patient 8	Bilateral cryptorchidism	nd
Patient 9 (Decipher 255792)	-	Diaphragmatic hernia
Patient 10 (Decipher 260107)	Cryptorchidism	
Patient 11 (Decipher 272645)	Bilateral cryptorchidism	nd
Patient 12 (Decipher 274063)	-	Subclinical hypothyroidism
Patient 13 (Decipher 284583)	-	-
Patient 14 (Decipher 272245)	Cryptorchidism, poorly developed scrotal sac	Transient splenomegaly
Patient 15 (Decipher 274751)	-	Oesophageal dysmotility
Patient 16 (Decipher 262739)	Unilateral cryptorchidism	-
Patient 17 (Decipher 279456)	-	-
Patient 18 (Decipher 270887)	Bilateral cryptorchidism	Obstipation
Patient 19 (Decipher 252324)	-	-
Patient 20	Cryptorchidism	-
Patient 21	-	-
Patient 22	-	-
Patient 23	-	-
Patient 24	Cryptorchidism	-
Patient 25	Unilateral cryptorchidism	-

	GTA	Other organ problems
PARIANI ET AL	Bilateral cryptorchidism	nd
HORN ET AL - Patient 1	nd	nd
HORN ET AL - Patient 2	nd	nd
HORN ET AL - Patient 3	nd	nd
HAMDAN ET AL - Patient 1	nd	nd
HAMDAN ET AL - Patient 2	nd	Atresia of the midjejunum and midileum gut
CARR ET AL	-	nd
O'ROAK ET AL	nd	nd
TALKOWSKI ET AL	nd	Spina bifida occulta, bilateral inguinal hernia
LE FEVRE ET AL	-	nd
PALUMBO ET AL	nd	nd
LOZANO ET AL	nd	Single umbilical hernia
SONG ET AL	nd	nd
TUTULAN-CUNITA ET AL *	Cryptorchidism, micropenis and scrotal abnormalities	nd
SOLLIS ET AL - Patient 1	nd	nd
SOLLIS ET AL - Patient 2	nd	nd
SOLLIS ET AL - Patient 3	nd	nd
THEVENON ET AL - Patient 1 (Decipher 257047)	nd	Partial growth hormone deficiency
THEVENON ET AL - Patient 2 (Decipher 2059)	nd	Intestinal malrotation
THEVENON ET AL - Patient 3 (Decipher 4438)	nd	nd
DIMITROV ET AL - Patient 1	nd	nd
DIMITROV ET AL - Patient 2	Bilateral cryptorchidism, micropenis and scrotal hypoplasia	Absence of gallbladder
DIMITROV ET AL- Patient 3	Hypoplastic labia majora	nd

Patient 1
Patient 2
Patient 3
Patient 4
Patient 5
Patient 6
Patient 7 (Decipher 331530)
Patient 8
Patient 9 (Decipher 255792)
Patient 10 (Decipher 260107)
Patient 11 (Decipher 272645)
Patient 12 (Decipher 274063)
Patient 13 (Decipher 284583)
Patient 14 (Decipher 272245)
Patient 15 (Decipher 274751)
Patient 16 (Decipher 262739)
Patient 17 (Decipher 279456)
Patient 18 (Decipher 270887)
Patient 19 (Decipher 252324)
Patient 20
Patient 21
Patient 22
Patient 23
Patient 24
Patient 25

Other relevant history

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Viral (enterovirus) meningitis at age 7 months. Recurrent otitis.
Deceased due to aspiration pneumonia at the age of 16 years.

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Deceased at age 5 months due to complications of diaphragmatic hernia.

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Recurrent fever of unknown origin. Urinary dribbling.
Father has macrocephaly (98th Centile).

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Mixed enuresis.

-

-

-

Incontinentia diurna.

-

-

Prematurity. Listeria infection. BPD.

-

PARIANI ET AL

HORN ET AL - Patient 1

HORN ET AL - Patient 2

HORN ET AL - Patient 3

HAMDAN ET AL - Patient 1

HAMDAN ET AL - Patient 2

CARR ET AL

O'ROAK ET AL

TALKOWSKI ET AL

LE FEVRE ET AL

PALUMBO ET AL

LOZANO ET AL

SONG ET AL

TUTULAN-CUNITA ET AL *

SOLLIS ET AL - Patient 1

SOLLIS ET AL - Patient 2

SOLLIS ET AL - Patient 3

THEVENON ET AL - Patient 1 (Decipher 257047)

THEVENON ET AL - Patient 2 (Decipher 2059)

THEVENON ET AL - Patient 3 (Decipher 4438)

DIMITROV ET AL - Patient 1

DIMITROV ET AL - Patient 2

DIMITROV ET AL - Patient 3

Other relevant history

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-

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Enuresis.

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Skin hypopigmentation.

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Enuresis

-

Deceased at the age of 3 years due to infectious event.

Deceased.

-

Pneumothorax and pulmonary hypertension at birth. Recurrent aspiration pneumonia. Chronic lung disease. Sleep apnea.

Profound respiratory distress and cyanosis at birth which required intubation. Deceased at age of 4 months.

Fetal distress and meconium aspiration requiring resuscitation and ventilation. Deceased at age of 2 years.