Patient	Cognitive Features	Growth Features	Facial Features	Skeletal Features	Heart Features	Neurologic Features	Other Features
#1 2q11.2 gain	Moderate developmental delay	Short stature	Midface hypoplasia with hypoplastic nose	Macrocephaly with frontal bossing and dolichocephaly; significant shortening of all segments of the extremities, significant brachydactyly	NFE	Hypotonia with continued head lag at age 15 months	None
#2 2q11.2 loss	Learning disabilities, ADHD	Normal growth parameters	U	Left lumbar scoliosis	Aortic coarctation requiring surgical repair	U	Café au lait spots
#3 2q13 gain	Severe developmental delay	At 3 years 1 month: all growth parameters <5th percentile	Prominent nose with bulbous nasal tip, unusual ear pit, dental crowding	Mild microcephaly with bitemporal narrowing, severe scoliosis, joint contractures involving hands, knees, and ankles	NFE	Congenital hypomyelinating neuropathy, hypotonia	Retinitis pigmentosa, bilateral cryptorchidism
#4 2q13 gain	Mild developmental delay, apraxia, ADHD, and anxiety	At 13 years 9 months: weight 5th-10th percentile; height 50th percentile; head circumference 10th percentile	Small jaw and dental crowding, hypertelorism, broad nasal bridge, small mouth with arched palate	5th digit clinobrachydactyly	Innocent heart murmur	Tourette syndrome	Primary IgG-2 subclass deficiency, mild unilateral sensorineural hearing loss
#5 2q13 loss	NA	U	U	U	Heterotaxy with multiple CHDs: TAPVR, VSD, PDA, coarctation of aorta	Sleep apnea, seizures	
#6	Severely impaired	U	Mild face retraction, widely	Broad feet with short toes	U	U	U

2q13 loss	speech and language skills		spaced teeth				
#7 2q13 loss	NA	IUGR	Dysmorphic facies: microcephaly, low-set ears, micrognathia, neck webbing	Rocker-bottom feet, wide fingers and toes with small/absent nails, clenched hands with distal finger contractures	VSD and PFO	Agenesis of the corpus callosum, apnea, hypotonia	Esophageal atresia, inguinal hernia, small penis
#8 7q11.21 loss	No indication provided	U	U	U	U	U	U
#9 7q36.1 loss	Mild developmental delay	Head circumference and weight in 5th percentile	Low-set, posteriorly rotated ears, widely spaced eyes	U	Normal cardiology evaluation with EKG	Generalized tonic-clonic seizures; normal head CT scan	U
#10 17q23 loss	Mild developmental delay, especially speech and language	Weight 10th percentile; height 25-50th percentile; head circumference 10th percentile	Frontal bossing, curly hair without coarseness, stellate pattern to irides, rounded nasal tip, gap between 1st and 2nd teeth	Long fingers and toes; 2nd toe overlaps 1st and 3rd toes on the R, 2nd and 4th toes overlap 3rd on the L	NFE	Hyperactivity, separation anxiety	None

"U" = information unavailable; "NFE" = not formally evaluated; "NA"= not able to asses due to young age; "CHD" = congenital heart defect; "TAPVR" = total anomalous pulmonary venous return; "VSD" = ventriculoseptal defect; "PDA" = patent ductus arteriosus; "IUGR" = intrauterine growth retardation; " PFO" = patent foramen ovale; "EKG" = electrocardiogram; "CT" = computed tomography; "GA" = gestational age; "R" = right; "L" = left

Supplemental Table 3. Phenotypic features of patients described in Table 1.