

Frequency of Medical and Developmental Problems in 18p-

	Incidence in 112 cases reported in the literature
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<i>NEONATAL COMPLICATIONS</i>	38 (34%)
Respiratory distress	12 (11%)
FTT/Feeding difficulties	14 (13%)
Jaundice	4 (4%)
Death within 1 yr	8 (7%)
Cyanosis	4 (4%)
Reported in single individuals	Lactose intolerance Neonatal sepsis Hypoglycemia Lymphedema
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<i>CARDIAC ANOMALIES^a</i>	12 (11%)
VSD	5 (4%)
Heterotaxy/dextrocardia	2 (2%)
Tetralogy of Fallot	2 (2%)
Pulmonic stenosis	3 (3%)
PDA	5 (4%)
Coarctation of aorta	2 (2%)
Reported in single individuals	Transposition of the great arteries Congenital calcification of the atrioventricular node
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<p>^a The majority of patients did not have an echocardiogram. Thus, it is possible that some patients had heart defects that were simply undetected.</p>	
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<i>GASTROINTESTINAL</i>	18 (16%)
Inguinal hernia	4 (4%)
Volvulus/Malrotation	2 (2%)
Splenic malformation (Fissured or accessory)	3 (3%)
HSM	3 (3%)
Dextroposition	2 (2%)
Dysphagia	2 (2%)
Reported in single individuals	Excessive drooling Congenital subglottic stenosis Diaphragmatic hernia Hiatal hernia Umbilical hernia Gastric carcinoma
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<i>ORTHOPEDIC ANOMALIES</i>	30 (27%)
Pes planus	5 (4%)
Scoliosis or kyphosis	6 (5%)
Cubitus valgus	4 (4%)
Pectus abnormalities	6 (5%)
Genu valgus	2 (2%)

Spina bifida occulta	2 (2%)
Bone age abnormalities	3 (3%)
Reported in single individuals	
	Tight ACL's
	Derotated femora
	Bilateral clubfoot
	Cervical ribs
	Skull deformity
	Congenital hip dysplasia
	Cervical spondylosis

NEUROLOGICAL ANOMALIES 46 (41%)

Hypotonia	24 (21%)
Dystonia	5 (4%)
HPE/HPE variant	13 (12%)
Hypertonia/spasticity/contractures	4 (4%)
Microcephaly	5 (4%)
Febrile seizures	2 (2%)
Reported in single individuals	
	Apathetic
	Parkinson-like signs
	Hemiplegia
	Hypokinesia
	Torticollis
	Hemiparesis
	Seizures

OPHTHALMOLOGIC ABNORMALITIES 39 (35%)

Strabismus	20 (18%)
Ptosis	21 (19%)
Hyperopia	2 (2%)
Myopia	4 (4%)
Astigmatism	3 (3%)
Nystagmus	3 (3%)
Exophthalmos	4 (4%)
Reported in single individuals	
	Glaucoma in late 30's
	Tiny pupils/small globes

CLEFT LIP/PALATE 24 (21%)

Midline cleft lip/Palate	3 (3%)
High-arched palate	12 (11%)
Unilateral CL/CP	2 (2%)
Cleft Palate	3 (3%)
Reported in single individuals	
	Bilateral cleft lip/palate
	Cleft Uvula
	Torus palatinus
	"Severe clefts"

GENITOURINARY ANOMALIES 18 (16%)

Small penis	6 (5%)
Uterine abnormalities	2 (2%)

Changes in labia majora	3 (3%)
Reported in single individuals	Hydrocele Cryptorchidism Recurrent UTI's Pyelonephritis Microalbuminuria Thamuria Hypoplastic left kidney Hypospadias

ENDOCRINOLOGY ANOMALIES 22 (20%)

Hypothyroidism	4 (4%)
GH deficiency	5 (4%)
Diabetes	2 (2%)
Reported in single individuals	Androgen insensitivity Pituitary absence Adrenal hypoplasia Gonadal dysgenesis Thyroid dysplasia Thyroiditis

OTOLOGIC ANOMALIES 8 (7%)

Recurrent otitis media	6 (5%)
Hearing loss	3 (3%)
Reported in single individuals	Choanal stenosis

HEAD IMAGING ABRNORMALITIES^b 28 of 37 (76%)

Holoprosencephaly	11 (30%) ^c
Pituitary abnormalities	6 (16%) ^c
Signal abnormalities	6 (16%) ^c
Ventricle dilation/abnormalities	8 (22%) ^c
Reported in single individuals	Dandy-Walker cyst Small hemorrhage in R lateral ventricle Multiple anomalies Cortico-subcortical atrophy Hypoplasia of the hypothalamus Thin corpus callosum

^b 37 of the 112 patients had head imaging studies.

^c Percentage of those who had head imaging studies

DEVELOPMENT^d 50 of 57 (88%)

Developmental Delays	46 (81%)
Speech delay ^e	30 (53%)
Normal intelligence	7 (12%)
Normal intelligence to borderline mental retardation	3 (5%)
Borderline to mild mental retardation	9 (16%)
Mild mental retardation	17 (30%)
Mild to moderate mental retardation	1 (2%)
Moderate mental retardation	7 (12%)

Moderate to severe mental retardation	2 (4%)
Severe mental retardation	1 (2%)
Profound mental retardation	1 (2%)
Mental retardation (degree not specified)	13 (23%)

^d Information regarding development was only available in 57 of the 112 case reports. The figures in this section represent the percentage of those 57 case reports that reported data on development.

^e Refers to patients that were explicitly reported to have significant speech delays

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