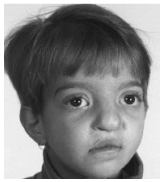
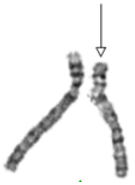
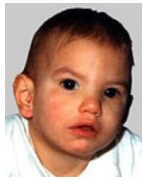
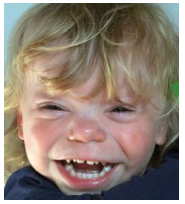
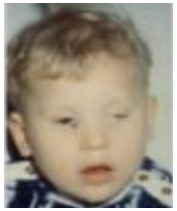
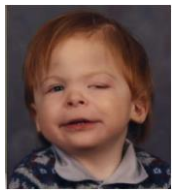
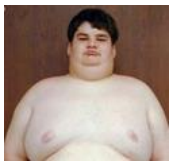
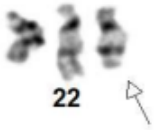
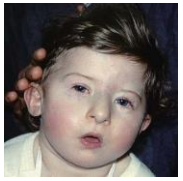
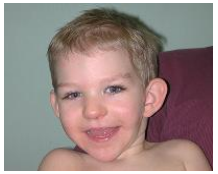
Deletion Syndrome	Chromosome Abnormality	Critical Gene(s)	Clinical Features		Chromosomes
1p36 deletion syndrome	del(1)(p36.3) *(terminal, breakpoints vary)*	Many candidate genes	Intellectual disability, behavior problems, structural brain abnormalities, hypotonia, dysphagia, microbrachycephaly, dysmorphic facies (deep-set eyes, straight eyebrows, midface hypoplasia, broad flat nose, long philtrum, pointed chin, low set ears), vision/hearing problems, skeletal/heart/GI/kidney/genital abnormalities		
Wolf-Hirschhorn syndrome	del(4)(p15.3) *(terminal, breakpoints vary; other mechanisms such as translocations also occur)*	*LETM1, WHSC1,* and others	Intellectual disability and growth deficiency, microcephaly, hypertelorism, broad nasal bridge, down-turned mouth, cleft lip and/or palate, micrognathia, cryptorchidism, hypospadias, "Greek Warrior helmet" appearance		
Cri-du-Chat syndrome	del(5)(p15.3) *(terminal, breakpoints vary; other mechanisms such as translocations also occur)*	Multiple critical regions have been described for different aspects of the phenotype	Intellectual disability and growth deficiency, cat-like cry in infancy, microcephaly, round face, hypertelorism, downlanting palpebral fissures		
Williams syndrome	del(7)(q11.23q11.23) *(interstitial)*	*ELN*	Intellectual disability, short stature, cardiovascular anomalies (most commonly SVAS or supravalvular aortic stenosis), friendly personality, hoarse voice, periorbital fullness, stellate pattern of the iris, anteverted nares, long philtrum, full lips		
Langer-Giedion syndrome	del(8)(q23.3q24.11) *(interstitial)*	*TRPS1, EXT1*	AKA Tricho-Rhino-Phalangeal syndrome Type II Intellectual disability and growth retardation, multiple exostoses, cone shaped epiphyses, fine scalp hair, bulbous nose, prominent ears, simple but prominent philtrum, loose redundant skin in infancy		
WAGR	del(11(p13) *(terminal)*	*WT1* , *PAX6*	Wilms tumor, aniridia, genitourinary anomalies (males- cryptorchidism, females- streak gonads, bicornate uterus), intellectual disability		

Jacobsen syndrome	del(11)(q24.1) *(terminal)*	*BSX, NRGN, ETS1* , *FLI1, RICS (ARHGAP32)*	Intellectual disability and growth retardation, trigonocephaly, strabismus, cardiac defects, digit anomalies, thrombocytopenia		
Pallister-Killian syndrome	mosaic +i(12)(p10)		Intellectual disability, hypotonia, limited/absent speech, coarse facial features (high rounded forehead, broad nasal bridge, widely spaced eyes, low-set ears, rounded cheeks, wide mouth, thin upper lip, large tongue), cleft palate, skeletal abnormalities, diaphragmatic hernia		
Angelman syndrome	del(15)(q11.2q13) involving maternal homolog *(interstitial)*	*UBE3A*	Intellectual disability and developmental delay, frequent laughter, ataxia and jerky movements, seizures, maxillary hypoplasia, deep-set eyes, large mouth with protruding tongue, widely spaced teeth, prognathia		
Prader-Willi syndrome	del(15)(q11.2q13) involving paternal homolog *(interstitial)*	*SNRPN*	Intellectual disability and developmental delay, hypotonia and feeding problems in infancy, later obesity associated with hyperphagia, narrow bifrontal diameter, almond-shaped eyes, small hands and feet, hypogonadism, skin picking		
Miller-Dieker syndrome	del(17)(p13.3) *(terminal; other mechanisms such as translocations also occur)*	*LIS1 (PAFAH1B1)* , *YWHAE*	Intellectual disability and developmental delay, lissencephaly, microcephaly, bitemporal depression, thin upper lip, micrognathia, ear dysplasia, anteverted nostrils		
Smith-Magenis syndrome	del(17)(p11.2p11.2) *(interstitial)*	*RAI1*	Intellectual disability, behavioral problems, hyperactivity, sleep disturbances, decreased pain sensitivity, short stature, brachycephaly, midface hypoplasia, prognathism, fingertip pads, hoarse voice		

Cat-Eye syndrome	+idic(22)(q11.2) *(typically a supernumerary chromosome, can also be seen as interstitial triplication)*	*CECR2, SLC25A18, ATP6V1E1*	Intellectual disability, coloboma, anal atresia, preauricular skin tags or pits, heart defects (total anomalous pulmonary venous return, among others), kidney/genital defects, skeletal defect		
22q11.2 deletion syndrome (velocardiofacial / DiGeorge syndrome)	del(22)(q11.2q11.2) *(interstitial)*	*TBX1*	Conotruncal cardiac defects (tetralogy of Fallot, interrupted aortic arch, truncus arteriosus, ventricular septal defect, among others), palate abnormalities (cleft palate, velopharyngeal incompetence), short stature, immunodeficiency, learning disabilities, psychiatric disorders, dysmorphic features (prominent nose, narrow palpebral fissures, elongated face and/or nose, thin lips)		
Phelan McDermid syndrome	del(22)(q13.3) *(terminal; other mechanisms such as translocations also occur)*	*SHANK3*	Intellectual disability, hypotonia, absent/delayed speech, autistic behaviors, decreased sensitivity to pain, reduced sweat, cyclic vomiting, gastroesophageal reflux, dysmorphic features (long narrow head, prominent ears, pointed chin, ptosis, deep set eyes, syndactyly, abnormal toenails), some with accelerated growth		

Chromosome images provided courtesy of Cincinnati Children's Hospital Medical Center Cytogenetics Laboratory

Additional image sources:
https://en.wikipedia.org/wiki/1p36_deletion_syndrome#
http://medgen.genetics.utah.edu/photographs/pages/wolfhirschhorn.htm
https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-1-33
https://healthjade.com/williams-syndrome/
https://www.facebook.com/NationwideChildrensHospital/photos/a.10153610596630135/10153610597175135/?type=3
https://quizlet.com/515379284/nicu-genetics-flash-cards/
https://ojrd.biomedcentral.com/articles/10.1186/1750-1172-4-9
https://positiveexposure.org/album/pallister-killian-syndrome/#
https://en.wikipedia.org/wiki/Angelman_syndrome
https://www.nature.com/articles/ejhg2008165
https://medlineplus.gov/genetics/condition/miller-dieker-syndrome/
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3152558/
https://www.slideshare.net/slideshow/genetics-12020491/12020491
https://en.wikipedia.org/wiki/DiGeorge_syndrome
https://www.spectrumnews.org/news/study-catalogs-features-of-phelan-mcdermid-syndrome/