Condition	Implicated gene	Hallmark S/Sx
Achondroplasia	mutations in FGFR3 gene	disproportionate short stature, megalencephaly, frontal bossing, midface hypoplasia, arm/leg shortening, prominent lumbar lordosis, genu varum, trident hand configuration
Angelman Syndrome	deletion of 15q11.2-13 (maternal gene)	frequent laughter or smiling, severe developmental delay, speech difficulties, seizure disorders
CHARGE Syndrome	Mutation of CHD7 gene	nonrandom association of coloboma, heart anomalies, choanal atresia, developmental retardation, genital/ear anomalies
Cockayne Syndrome	mutations in ERCC6 gene or	premature aging, cachectic dwarfism, thinning of skin/hair, sunken eyes, stooped posture
Cornelia De Lange Syndrome	mutation of NIPBL, among others	distinctive facial appearance, prenatal/postnatal growth deficiency, feeding difficulties, psychomotor delay, behavioral problems
Cri-du-chat Syndrome	deletion of a B group chromosome (Bp-), later identified as 5p-	high-pitched "cat" cry in infancy w/ FTT, microcephaly, facial abnormalities/laxity, MR
Crouzon Syndrome	mutations in FGFR2	craniofacial dysostosis, premature closure sutures, ypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, hypoplastic maxilla, and relative mandibular prognathism
Danon Disease	Lysosomal-associated membrane protein-2 (LAMP2)	rare form of HCM and muscular dystrophy
Down Syndrome	trisomy of chromosome 21	flat occiput, low-set ears, small nose w/ depressed nasal bridge, protruding tongue, high-arch palate, dental abnormalities
Ellis-van Creveld Syndrome	unknown	Disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, small chest, congenital heart defects
Fragile X Syndrome (Marvin Bell Syndrome)	unidentified mode of X-linked inheritance, perhaps excess long-arm extension	cognitive, behavioral, and neuropsychological difficulties, autistic-like behaviors, ADHD, MR, shy, sensory integration issues
Kearns-Sayre Syndrome	linked to mtDNA	ophthalmoparesis/pigmentary retinopathy < 20 yo, short stature, endocrinopathies, bilat sensorineural hearing loss
Klinefelter Syndrome	extra sex chromosome (genotype XXY)	hypogonadism (small testes, azoospermia, oligospermia), gynecomastia in late puberty, psychosocial problems, hyalinization/fibrosis of the seminiferous tubules, elevated urinary gonadotropin levels
Marfan Syndrome	mutations in FBN1 gene on chromosome 15	tall stature, ectopia lentis, mitral valve prolapse, aortic root dilatation, and aortic dissection
Neurofibromatosis (von Recklinghausen disease)	mutation/deletion of NF1 gene	development of neurofibromas, freckling in groin/axillary, >6 café-au-lait spots, Lisch nodules, bowing in legs, optic nerve tumors
Noonan Syndrome	Mutations in the RAS-MAPK signaling pathway	unusual facies (ie, hypertelorism, down-slanting eyes, webbed neck), congenital heart disease (in 50%), short stature, and chest deformity
Osteogenesis Imperfecta	mutations in the genes that codify for type I procollagen (ie, COL1A1 andCOL1A2)	multiple fractures, bruise easily, deafness

Patau Syndrome	extra copy of chromosome 13	severe mental deficiency, holoprosencephaly, polydactyly, flexion of the fingers, rocker-bottom feet, facial clefting, neural tube defects, heart defects
Prater Willi	deletion of 15q11.2-13 (paternal gene)	diminished fetal activity, obesity, hypotonia, MR, short stature, hypogonadotropic hypogonadism, strabismus, small hands/feet
Proteus Syndrome	unknown, mosiacism	Partial gigantism with limb or digital overgrowth, unusual body habitus, cerebriform thickening of feet sole, cutaneous and subcutaneous lesions
Rubinstein-Taybi Syndrome	deletions in band 16p13, mutations in the EP300 gene	distinctive facies, mental retardation, broad thumbs, and broad great toes
Silver-Russell Syndrome	hypomethylation of chromosome 11p15, +/- maternal uniparental disomy of chromosome 7	LBW, developmental delay, increased sweating in infancy, feeding difficulties, motor impairment
Sjogren-Larsson Syndrome	inborn error of lipid metabolism	MR, spastic diplegia/tetraplegia, seizures, photophobia
Treacher Collins Syndrome	unknown	facial dysmorphology, kyphotic cranial base, short palpebral fissures sloped laterally downward, pinna abnormalities, cleft palate, etc.
Trisomy 18	trisomy of chromosome 18	severe psychomotor and growth retardation, microcephaly, microphthalmia, malformed ears, micrognathia/retrognathia, microstomia, clenched fingers, congenital malformations
Tuberous Sclerosis	Mutations in either of 2 genes (TSC1 and TSC2)	MR, epilepsy, facial angiofibromas, cortical tubers, subependymal nodule, subependymal giant cell astrocytomas, facial angiofibromas, hypomelanotic spots known as Fitzpatrick patches (ash-leaf spots), cardiac rhabdomyomas, renal angiomyolipomas
Turner Syndrome	absence of one set of genes from the short arm of one X chromosome	short stature, ovarian failure, high-arch palate, shield chest, abnormal hair line, webbed neck, pedal edema at birth
van der Woude Syndrome	deletion in chromosome 1q32-q41; 1p34 also implicated	acleft lip or cleft palate, pits of lips (autosomal dominant)
Waardenburg Syndrome	mutations in PAX3	hearing loss, dystopia canthorum, and pigmentary abnormalities of the hair, skin, and eyes
Wolf-Hirschhorn Syndrome	deletion of the distal short arm of chromosome 4	delayed psychomotor, seizures, speech abnormalities, stereotypies, microcephaly, frontal bossing, dysmorphic facial features, beaked nose
Wolff-Parkinson-White Syndrome	mutation in PRKAG2 gene	