

2018 Etiologies in Order of Frequency by Category

Hereditary Syndromes and Disorders	Count
CHARGE Syndrome	958
Down syndrome (Trisomy 21 syndrome)	308
Usher I syndrome	252
Stickler syndrome	130
Dandy Walker syndrome	119
Cornelia de Lange	102
Goldenhar syndrome	98
Usher II syndrome	83
Wolf-Hirschhorn syndrome (Trisomy 4p)	68
Trisomy 13 (Trisomy 13-15, Patau syndrome)	60
Pierre-Robin syndrome	57
Moebius syndrome	55
Trisomy 18 (Edwards syndrome)	52
Norrie disease	38
Leber congenital amaurosis	35
Chromosome 18, Ring 18	31
Aicardi syndrome	29
Alstrom syndrome	27
Pfieffer syndrome	27
Treacher Collins syndrome	27
Waardenburg syndrome	27
Marshall syndrome	25
Refsum syndrome	21
Cri du chat syndrome (Chromosome 5p- synd)	16
Bardet-Biedl syndrome (Laurence Moon-Biedl)	15
Hurler syndrome (MPS I-H)	15
Crouzon syndrome (Craniofacial Dysotosis)	13
NF1 - Neurofibromatosis (von Recklinghausen dis)	13
Kniest Dysplasia	12
Turner syndrome	11
Usher III syndrome	10
Cockayne syndrome	9
Apert syndrome/Acrocephalosyndactyly, Type 1	8
Leigh Disease	8
Alport syndrome	6
Monosomy 10p	6
NF2 - Bilateral Acoustic Neurofibromatosis	6
Batten disease	5
Kearns-Sayre syndrome	5
Klippel-Feil sequence	5

Hereditary Syndromes and Disorders	Count
Prader-Willi	5
Sturge-Weber syndrome	5
Marfan syndrome	3
Hand-Schuller-Christian (Histiocytosis X)	2
Hunter Syndrome (MPS II)	2
Maroteaux-Lamy syndrome (MPS VI)	2
Morquio syndrome (MPS IV-B)	2
Optico-Cochleo-Dentate Degeneration	2
Smith-Lemli-Opitz (SLO) syndrome	2
Wildervanck syndrome	2
Herpes-Zoster (or Hunt)	1
Vogt-Koyanagi-Harada syndrome	1
OTHER: Hereditary Syndromes/ Disorders	1855

Pre Natal Congenital Complications	Count
Cytomegalovirus (CMV)	293
Microcephaly	211
Hydrocephaly	203
Maternal Drug Use	71
Congenital Rubella	33
Fetal Alcohol syndrome	17
Congenital Toxoplasmosis	11
Neonatal Herpes Simplex (HSV)	8
Congenital Syphilis	2
OTHER: Pre-Natal/ Congenital Complications	490

Post Natal Non-Congenital Complications	Count
Asphyxia	184
Severe Head Injury	137
Meningitis	132
Tumors	91
Stroke	57
Encephalitis	49
Infections	30
Chemically Induced	16
Direct Trauma to the eye and/or ear	12
OTHER: Post-Natal/ Non-Congenital	437

Additional	Count
Complication of Prematurity	991
No Determination of Etiology	1745