

2017 Etiologies in Order of Frequency by Category

Hereditary Syndromes and Disorders

CHARGE Syndrome	933
Down syndrome (Trisomy 21 syndrome)	315
Usher I syndrome	233
Stickler syndrome	137
Dandy Walker syndrome	115
Goldenhar syndrome	102
Cornelia de Lange	99
Usher II syndrome	85
Wolf-Hirschhorn syndrome (Trisomy 4p)	72
Trisomy 13 (Trisomy 13-15, Patau syndrome)	61
Moebius syndrome	50
Pierre-Robin syndrome	48
Trisomy 18 (Edwards syndrome)	47
Norrie disease	37
Leber congenital amaurosis	36
Aicardi syndrome	34
Chromosome 18, Ring 18	31
Waardenburg syndrome	31
Pfieffer syndrome	27
Marshall syndrome	26
Alstrom syndrome	25
Treacher Collins syndrome	24
Cri du chat syndrome (Chromosome 5p- synd)	23
Hurler syndrome (MPS I-H)	19
Refsum syndrome	18
Bardet-Biedl syndrome (Laurence Moon-Biedl)	15
Crouzon syndrome (Craniofacial Dysotosis)	15
Cockayne syndrome	13
NF1 - Neurofibromatosis (von Recklinghausen dis)	13
Usher III syndrome	12
Kniest Dysplasia	11
Turner syndrome	9
Sturge-Weber syndrome	8
Apert syndrome/Acrocephalosyndactyly, Type 1	7
Kearns-Sayre syndrome	7
NF2 - Bilateral Acoustic Neurofibromatosis	7
Batten disease	6
Monosomy 10p	6
Leigh Disease	5
Prader-Willi	4

Hereditary Syndromes and Disorders

Alport syndrome	3
Hunter Syndrome (MPS II)	3
Klippel-Feil sequence	3
Marfan syndrome	2
Morquio syndrome (MPS IV-B)	2
Optico-Cochleo-Dentate Degeneration	2
Smith-Lemli-Opitz (SLO) syndrome	2
Wildervanck syndrome	2
Hand-Schuller-Christian (Histiocytosis X)	1
Hallgren syndrome	1
Herpes-Zoster (or Hunt)	1
Maroteaux-Lamy syndrome (MPS VI)	1
OTHER: Hereditary Syndromes/ Disorders	1693

Pre Natal Congenital Complications

Cytomegalovirus (CMV)	293
Microcephaly	214
Hydrocephaly	206
Maternal Drug Use	85
Congenital Rubella	41
Fetal Alcohol syndrome	18
Congenital Toxoplasmosis	12
Neonatal Herpes Simplex (HSV)	7
Congenital Syphilis	1
OTHER: Pre-Natal/ Congenital Complications	514

Post Natal Non-Congenital Complications

Asphyxia	194
Severe Head Injury	160
Meningitis	136
Tumors	94
Stroke	56
Encephalitis	45
Infections	33
Direct Trauma to the eye and/or ear	14
Chemically Induced	14
OTHER: Post-Natal/ Non-Congenital	415

Complication of Prematurity	1047
No Determination of Etiology	1919