

Table: Identified primary aetiologies that may cause deafblindness

Aetiology	Aetiology
Hereditary/Chromosomal Syndromes and Disorders	
101 Aicardi syndrome	132 Moebius syndrome
102 Alport syndrome	133 Monosomy 10p
103 Alstrom syndrome	134 Morquio syndrome (MPS IV-B)
104 Apert syndrome (Acrocephalosyndactyly, Type 1)	135 NF1 - Neurofibromatosis (von Recklinghausen disease)
105 Bardet-Biedl syndrome (Laurence Moon-Biedl)	136 NF2 - Bilateral Acoustic Neurofibromatosis
106 Batten disease	137 Norrie disease
107 CHARGE Syndrome	138 Optico-Cochleo-Dentate Degeneration
108 Chromosome 18, Ring 18	139 Pfeiffer syndrome
109 Cockayne syndrome	140 Prader-Willi
110 Cogan Syndrome	141 Pierre-Robin syndrome
111 Cornelia de Lange	142 Refsum syndrome
112 Cri du chat syndrome (Chromosome 5p- syndrome)	143 Scheie syndrome (MPS I-S)
113 Crigler-Najjar syndrome	144 Smith-Lemli-Opitz (SLO) syndrome
114 Crouzon syndrome (Craniofacial Dysostosis)	145 Stickler syndrome
115 Dandy Walker syndrome	146 Sturge-Weber syndrome
116 Down syndrome (Trisomy 21 syndrome)	147 Treacher Collins syndrome
117 Goldenhar syndrome	148 Trisomy 13 (Trisomy 13-15, Patau syndrome)
118 Hand-Schuller-Christian (Histiocytosis X)	149 Trisomy 18 (Edwards syndrome)
119 Hallgren syndrome	150 Turner syndrome
120 Herpes-Zoster (or Hunt)	151 Usher I syndrome
121 Hunter Syndrome (MPS II)	152 Usher II syndrome
122 Hurler syndrome (MPS I-H)	153 Usher III syndrome
123 Kearns-Sayre syndrome	154 Vogt-Koyanagi-Harada syndrome
124 Klippel-Feil sequence	155 Waardenburg syndrome
125 Klippel-Trenaunay-Weber syndrome	156 Wildervanck syndrome
126 Kniest Dysplasia	157 Wolf-Hirschhorn syndrome (Trisomy 4p)
127 Leber congenital amaurosis	158 Rosenberg Chutorian Syndrome
128 Leigh Disease	159 Wolfram Syndrome (DIDMOAD)
129 Marfan syndrome	160 Zelweger Syndrome (Cerebrohepatorenal Syndrome)
130 Marshall syndrome	199 Other
131 Maroteaux-Lamy syndrome (MPS VI)	

Pre-Natal/Congenital Complications	Post-Natal/Non-Congenital Complications
201 Congenital Rubella	301 Asphyxia
202 Congenital Syphilis	302 Direct Trauma to the eye and/or ear
203 Congenital Toxoplasmosis	303 Encephalitis
204 Cytomegalovirus (CMV)	304 Infections
205 Foetal Alcohol syndrome	305 Meningitis
206 Hydrocephaly	306 Severe Head Injury
207 Maternal Drug Use	307 Stroke
208 Microcephaly	308 Tumours
209 Neonatal Herpes Simplex (HSV)	309 Chemically Induced
299 Other	399 Other
Related to Prematurity	Undiagnosed
401 Complications of Prematurity	501 No Determination of Aetiology