

## **Hereditary/Chromosomal Syndromes and Disorders**

**(# FAVI constituents recorded)**

Aicardi syndrome  
Alport syndrome  
Alstrom syndrome  
Apert syndrome (Acrocephalosyndactyly, Type 1)  
Bardet-Biedl syndrome (Laurence Moon-Biedl)  
Batten disease  
**CHARGE syndrome (44)**  
Chromosome 18, Ring 18  
Cockayne syndrome  
Cogan Syndrome  
**Cornelia de Lange (2)**  
Cri du chat syndrome (Chromosome 5p-syndrome)  
Crigler-Najjar syndrome  
Crouzon syndrome (Craniofacial Dysostosis)  
**Dandy Walker syndrome (2)**  
**Down syndrome (Trisomy 21 syndrome) (14)**  
**Goldenhar syndrome (8)**  
Hand-Schuller-Christian (Histiocytosis X)  
Hallgren syndrome  
Herpes-Zoster (or Hunt)  
Hunter Syndrome (MPS II)  
**Hurler syndrome (MPS I-H) (1)**  
Kearns-Sayre syndrome  
Klippel-Feil sequence  
Klippel-Trenaunay-Weber syndrome  
Kniest Dysplasia  
**Leber congenital amaurosis (1)**  
Leigh Disease  
Marfan syndrome

Complications of Prematurity (65)  
No Determination of Etiology (102)

Marshall syndrome  
Maroteaux-Lamy syndrome (MPS VI)  
**Moebius syndrome (1)**  
**Monosomy 10p (1)**  
Morquio syndrome (MPS IV-B)  
**NF1 - Neurofibromatosis (von Recklinghausen disease) (1)**  
NF2 - Bilateral Acoustic Neurofibromatosis  
**Norrie disease (2)**  
Optico-Cochleo-Dentate Degeneration  
**Pfieffer syndrome (3)**  
Prader-Willi  
Pierre-Robin syndrome  
Refsum syndrome  
Scheie syndrome (MPS I-S)  
Smith-Lemli-Opitz (SLO) syndrome  
**Stickler syndrome (8)**  
Sturge-Weber syndrome  
**Treacher Collins syndrome (3)**  
**Trisomy 13 (Trisomy 13-15, Patau syndrome) (5)**  
**Trisomy 18 (Edwards syndrome) (2)**  
**Turner syndrome (1)**  
**Usher I syndrome (11)**  
**Usher II syndrome (3)**  
**Usher III syndrome (1)**  
Vogt-Koyanagi-Harada syndrome  
**Waardenburg syndrome (1)**  
Wildervanck syndrome  
**Wolf-Hirschhorn syndrome (Trisomy 4p) (1)**  
Other Hereditary/Chromosomal (37):  
Duane's syndrome, Frasier's syndrome, 13q deletion, Trisomy 9, 13, Aicardi-Goutieres syndrome 4, 1P 36 deletion syndrome, 4p deletion syndrome, partial trisomy 10, Gomez-Lopez-Hernandez syndrome, Peters anomaly, Steven Johnson syndrome, Pallister-Killian syndrome, Warfarin syndrome, DeMorsier's syndrome