

New GeneReviews (December 2008 – January 2009)

- CDC73-Related Disorders
- Congenital Hepatic Fibrosis Overview
- Primary Hyperoxaluria Type 2
- Congenital Stromal Corneal Dystrophy
- MYH9-Related Disorders

Newly Available Laboratory Tests in the United States

- 3-Methylglutaconic Aciduria Type 3
- ABCA4-Related Retinitis Pigmentosa
- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy 5
- Charcot-Marie-Tooth Neuropathy, Dominant Intermediate B
- Cone-Rod Dystrophy, Type 3
- Familial Exudative Vitreoretinopathy, Autosomal Dominant
- INS-Related Maturity-Onset Diabetes of the Young Type I
- Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
- Maternal Uniparental Disomy, Chromosome 14
- Osteoporosis Pseudoglioma Syndrome
- Paternal Uniparental Disomy, Chromosome 14
- Peters Anomaly
- Simpson-Golabi-Behmel Syndrome
- Stargardt Disease 1

Newly Available Laboratory Tests Internationally

Germany

- 2,4-Dienoyl-CoA Reductase Deficiency
- 3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency
- Biotinidase Deficiency

- Congenital Disorder of Glycosylation Im
- Congenital Disorder of Glycosylation In
- Congenital Disorder of Glycosylation IIc
- Congenital Disorder of Glycosylation IID
- Congenital Disorder of Glycosylation IIIf
- Dystonia 16
- Factor V Deficiency
- Factor X Deficiency
- Familial Paroxysmal Nonkinesigenic Dyskinesia
- Galactose Epimerase Deficiency
- Hartnup Disease
- Isovaleric Acidemia
- KCNE3-Related Hypokalemic Periodic Paralysis Recessive, TCell -Negative, B
- Ketothiolase Deficiency
- Kufor-Rakeb Syndrome
- PLOSL
- Phosphoglycerate Dehydrogenase Deficiency
- Renal-Hepatic-Pancreatic Dysplasia
- cblD
- cblD (variant 1)
- cblD (variant 2)