

New GeneReviews (January - February 2010)

- Autosomal Dominant Hyper IgE Syndrome
- Episodic Ataxia Type 1
- Mucopolipidosis III Gamma
- 17q21.31 Microdeletion Syndrome
- ALK-Related Neuroblastoma Susceptibility

Newly Available Laboratory Tests in the United States

- ABCC8-Related Transient Neonatal Diabetes Mellitus 2
- ACTN2-Related Familial Hypertrophic Cardiomyopathy
- AGRN-Related Congenital Myasthenic Syndrome
- ANKRD1-Related Dilated Cardiomyopathy
- ARL13B-Related Joubert Syndrome
- DSP-Related Ectodermal Dysplasia/Skin Fragility Syndrome
- Duane Retraction Syndrome 2
- ELN-Related Cutis Laxa
- Epidermolysis Bullosa, Lethal Acantholytic
- Familial Hemophagocytic Lymphohistiocytosis 5
- INPP5E-Related Joubert Syndrome
- Kallmann Syndrome 5
- LAMP2-Related Familial Hypertrophic Cardiomyopathy
- Limb Girdle Muscular Dystrophy Type 2L
- Long QT Syndrome 11
- Long QT Syndrome 12
- MASP2 Deficiency
- Mental Retardation, Autosomal Dominant 5
- Mitochondrial Respiratory Chain Complex IV Deficiency

- MKS1-Related Bardet-Biedl Syndrome
- Neurodegeneration due to Cerebral Folate Transport Deficiency
- Parkinsonism-Dystonia, Infantile
- Proprotein Convertase-1 Deficiency
- RASA1-Related Disorders
- RPGRIP1L-Related Joubert Syndrome
- PRPH2-Related Cone-Rod Dystrophy
- SLC6A5-Related Hyperekplexia
- Supravalvular Aortic Stenosis
- TMEM216-Related Joubert Syndrome
- TNNC1-Related Dilated Cardiomyopathy

Newly Available Laboratory Tests Internationally

France

- ABCC8-Related Transient Neonatal Diabetes Mellitus 2

Germany

- Charcot-Marie-Tooth Neuropathy Type 2B2
- Chorea-Acanthocytosis
- X-linked Mental Retardation, Nonspecific

Netherlands

- Hennekam Lymphangiectasia-Lymphedema Syndrome
- RASA1-Related Disorders
- Sick Sinus Syndrome 2, Autosomal Dominant
- Triosephosphate Isomerase Deficiency

Slovakia

- Budd-Chiari Syndrome

Sweden

- Ovarian Dysgenesis 2