

## New GeneReviews (November - December 2009)

- Thrombocytopenia Absent Radius Syndrome
- CATSPER-Related Male Infertility
- Dyskeratosis Congenita
- Tourette Disorder Overview

## Newly Available Laboratory Tests in the United States

- Acyl-CoA Dehydrogenase 9 Deficiency
- Atrial Septal Defect 5
- Childhood Restrictive Cardiomyopathy
- CHRNA1-Related Multiple Pterygium Syndrome Lethal Type
- CHRND-Related Multiple Pterygium Syndrome, Lethal Type
- Cone Dystrophy 3
- Cone-Rod Dystrophy, Type 12
- Cone-Rod Dystrophy, Type 5
- Cone-Rod Dystrophy, Type 7
- DFNB10 Nonsyndromic Hearing Loss and Deafness
- DFNB23 Nonsyndromic Hearing Loss and Deafness
- DFNB31 Nonsyndromic Hearing Loss and Deafness
- DFNB37 Nonsyndromic Hearing Loss and Deafness
- DFNB59 Nonsyndromic Hearing Loss and Deafness
- DOK7-Related Fetal Akinesia Deformation Sequence
- EPCAM-Related Hereditary Non-Polyposis Colon Cancer
- EYS-Related Retinitis Pigmentosa
- HPE8-Related Holoprosencephaly
- KLHL7-Related Retinitis Pigmentosa
- Laing Distal Myopathy
- Mental Retardation, X-linked, Syndromic 14
- MUSK-Related Congenital Myasthenic Syndrome
- MYH7-Related Myosin Storage Myopathy
- Nablus Mask-Like Facial Syndrome

- Noonan-Like Syndrome with Loose Anagen Hair/Omodysplasia 1
- PRPH2-Related Cone-Rod Dystrophy
- Scapuloperoneal Myopathy, MYH7-Related
- SNRNP200-Related Retinitis Pigmentosa
- UNC119-Related Cone-Rod Dystrophy
- Usher Syndrome Type 1G
- Usher Syndrome Type 2C
- Usher Syndrome Type 2D
- X-Linked Mental Retardation 21
- X-linked Mental Retardation 59
- ZDHHC9-Related X-linked Mental Retardation

## Newly Available Laboratory Tests Internationally

### Denmark

- Brugada Syndrome 5

### Germany

- EEM Syndrome
- HTRA2-Related Parkinson Disease
- Hypogonadotropic Hypogonadism
- Liver Failure, Acute Infantile
- Mitochondrial Respiratory Chain Complex II Deficiency, SDHAF1-Related
- Parkinson Disease, Susceptibility to Spinal Muscular Atrophy, X-Linked Infantile

### Italy

- Episodic Ataxia Type 5
- Episodic Ataxia Type 6

### Portugal

- 15q24 Microdeletion Syndrome
- 9q22.3 Microdeletion Syndrome

### Spain

- MMADHC-Related Methylmalonic Acidemia

### United Kingdom

- CFL2-Related Nemaline Myopathy
- Myotubular Myopathy, Recessive
- Spinal Muscular Atrophy, X-Linked Infantile
- Hypogonadotropic Hypogonadism