# new at GeneTests

*Full details of all of the information listed here can be found at www.genetests.org.*

## New *GeneReviews* (June-July 2008)

- Celiac Disease
- MED12-Related Disorders
- Infantile Neuroaxonal Dystrophy
- Hereditary Folate Malabsorption
- LMNA-Related Dilated Cardiomyopathy
- 46,XY Disorder of Sex Development and 46,XY Complete Gonadal Dysgenesis
- Hereditary Paraganglioma-Pheochromocytoma Syndromes
- Focal Dermal Hypoplasia

## Newly Available Laboratory Tests in the United States

- Alzheimer Disease Type 4
- Ataxia with Vitamin E Deficiency
- EIF2B1/EIF2B2/EIF2B3/EIF2B4-Related Childhood Ataxia with Central Nervous System Hypomyelination/Vanishing Whit
- Episodic Ataxia Type 1
- Fatal Infantile Lactic Acidosis
- Focal Segmental Glomerulosclerosis 2
- GABRG2-Related Generalized Epilepsy with Febrile Seizures Plus
- ICHTHYIN-Related Autosomal Recessive Congenital Ichthyosis
- Juvenile Myoclonic Epilepsy
- Mental Retardation, X-Linked, with Short Stature, Small Testes, Muscle Wasting, and Tremor
- Neurohypophyseal Diabetes Insipidus
- PRPF 3/PRPF 8/PRPF31/RP10-Related Retinitis Pigmentosa
- SEPN1-Related Congenital Fiber-Type Disproportion
- Spastic Paraplegia 8
- TPM3-Related Congenital Fiber-Type Disproportion

## Newly Available Laboratory Tests Internationally

### Norway

- ABCA1-Associated Familial High Density Lipoprotein Deficiency
- ABCA1-Related Disorders
- ACTC1-Related Dilated Cardiomyopathy
- APOA1-Associated Familial High Density Lipoprotein Deficiency
- Familial Hypercholesterolemia, Autosomal Dominant, 3
- Hyperalphalipoproteinemia
- Lecithin Cholesterol Acyltransferase Deficiency
- RP10-Related Retinitis Pigmentosa
- Severe Combined Immune Deficiency, Autosomal Recessive, T Cell-Negative, B Cell –Positive

### Germany

- ACTC1-Related Dilated Cardiomyopathy
- ACTC1-Related Familial Hypertrophic Cardiomyopathy
- ADAMTS10-Related Weill-Marchesani Syndrome
- CNGA1-Related Retinitis Pigmentosa
- CNGB1-Related Retinitis Pigmentosa
- COL11A1/COL11A2/COL2A1/COL9A1-Related Stickler Syndrome
- DFNA 8/12-/DFNB 3-/DFNB 6-/DFNB 7/11-/DFNB16-/DFNB18- Nonsyndromic Hearing Loss and Deafness
- Distal Hereditary Motor Neuronopathy Type VIIB
- FBN1-Related Weill-Marchesani Syndrome
- LGMD1A
- Limb-Girdle Muscular Dystrophy Type 2H, Type 2J and MERTK-Related Retinitis Pigmentosa
- Oguchi Disease
- PDE6A-/PDE6B-/PRPF31-Related Retinitis Pigmentosa
- Papillary Renal Carcinoma
- RGR-Related Retinitis Pigmentosa
- Retinal Dystrophy, Early-Onset, Severe
- Spastic Paraplegia 15
- Spinocerebellar Ataxia Type 11
- TARDBP-Related Amyotrophic Lateral Sclerosis
- TTN-Related Dilated Cardiomyopathy
- TTN-Related Familial Hypertrophic Cardiomyopathy
- Telethoninopathy
- USH2A-Related Retinitis Pigmentosa
- VAPB-Related Amyotrophic Lateral Sclerosis
- Vesicoureteral Reflux 2

### Canada

- Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy 5
- CTSD-/ MFSD8-Related Neuronal Ceroid-Lipofuscinosis

### United Kingdom

- Dysalbuminemic Hyperthyroxinemia

### Netherlands

- Hyper IgE Syndrome