new at GeneTests

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

The pace of change in medical genetics is rapid and continues to accelerate. GeneTests (www.genetests.org) has been an invaluable resource for the practicing geneticist seeking up-to-date information about genetic diseases, including the most recent genetic tests available. The GeneTests site also features a useful international directory of genetics clinics.

Here we list the newest Gene*Reviews* that can be found online at www.genetests.org. We also list newly available tests for genetic disorders (though readers are advised to check with genetests.org and the individual labs to be sure that the test is still available).

New Gene*Reviews* (July - August 2007)

- Baller-Gerold Syndrome
- CHMP2B-Related Frontotemporal Dementia
- Craniometaphyseal Dysplasia
- Dense Deposit Disease / Membranoproliferative Glomerulonephritis Type II
- Dilated Cardiomyopathy Overview
- Glycogen Storage Disease Type II (Pompe Disease)
- Hyperekplexia
- Optic Atrophy Type
- Tetra-Amelia Syndrome

Newly Available Laboratory Tests in the United States:

- 17-alpha-Hydroxylase-Deficient Congenital Adrenal Hyperplasia
- Ashkenazi Jewish Carrier Test Panels
- Cholesterol Desmolase-Deficient Congenital Adrenal Hyperplasia
- Chondrodysplasia Punctata 1, X-Linked Recessive
- Focal Dermal Hypoplasia
- GCK-Related Diabetes Mellitus, Permanent Neonatal
- Idiopathic Pulmonary Fibrosis
- Juvenile Polyposis/Hereditary Hemorrhagic Telangiectasia Syndrome
- Pseudo-Von Willebrand Disease
- RAF1-Related LEOPARD Syndrome
- RAF1-Related Noonan Syndrome
- SCN1B-Related Generalized Epilepsy with Febrile Seizures Plus

Newly Available Laboratory Tests Internationally

Belgium

- Berardinelli-Seip Congenital Lipodystrophy Type 1

Netherlands

- Autosomal Dominant Partial Epilepsy with Auditory Features
- GABRG2-Related Generalized Epilepsy with Febrile Seizures Plus
- Peters Plus Syndrome
- SCN2A-Related Febrile Seizures Associated with Afebrile Seizures

Turkey

- Pulmonary Alveolar Microlithiasis

United Kingdom

- SEPN1-Related Multiminicore Disease