Resources for Providers

**Medical Genetics in Pediatric Practice: A Handbook**
Embodying current Policy of the AAP, this new resource provides practice-focused help for addressing virtually any genetics-related issue you’re likely to confront. It’s replete with expert insights, pediatric-specific solutions, and quick-access aids you won’t find anywhere else. Consult this one-stop problem-solver for genetic processes, inheritance patterns, and genetic testing, summaries of common genetic disorders, images of anomalies that may indicate genetic conditions, case-based examples of ethical issues, and more.

**PediaGene: AAP Genetics Screening Guide: A mobile device application**
Based on the AAP manual Medical Genetics in Pediatric Practice, PediaGene is a one-stop resource for a wealth of genetic screening information, available in either patient encounter or reference context. This mobile device application is available for Windows and MAC operating systems.

**Online Mendelian Inheritance in Man (OMIM)**
This is a good source for finding information about individuals who have more than one congenital anomaly or a combination of unusual facial features, cognitive delays and/or a birth defect. Locating the conditions that list all of the features input for the search will help providers to narrow down the diagnosis. The Web site is most useful when two to three of the patient’s most unusual clinical features are entered into the Web site’s search engine. Too much non-specific information may not locate the right diagnosis and too little non-specific information may uncover too many conditions.

**Gene Tests/Gene Reviews** ([www.geneclinics.org](http://www.geneclinics.org))
Peer-reviewed disease descriptions written by experts. The Web site also includes an international directory of nearly 1,100 genetic and prenatal diagnostic clinics, an international directory of more than 610 genetic testing laboratories, and educational materials on more than 2,900 diseases. GeneReviews include a summary, clinical diagnostic information, diagnostic criteria, molecular genetic testing with pick-up rates and links to laboratories, testing strategies, detailed clinical information and natural history, differential diagnoses, disease management, registries, genetic counseling, and resources for families.

**AAP Committee on Genetics** ([http://www.aap.org/visit/cmte18.htm](http://www.aap.org/visit/cmte18.htm))
This resource includes physician-friendly health supervision guidelines which provide prenatal advice, recommended screening protocols and some disorder-specific growth curves. The Web site also contains newborn screening fact sheets, information about congenital adrenal hyperplasia, and guidelines on the clinical evaluation of children with mental retardation or developmental delays. Guidelines are available for common genetic topics:
- [Achondroplasia](http://www.aap.org/visit/cmte18.htm)
- [Down Syndrome](http://www.aap.org/visit/cmte18.htm)
• **Fragile X**
• **Marfan Syndrome**
• **Neurofibromatosis**
• **Prader-Willi Syndrome**
• **Sickle Cell Disease**
• **Turner Syndrome**
• **Williams Syndrome**


This Web site hosted by Medscape contains quality pictures and radiographs. Although not specific to genetics, the Web site includes summaries of many genetic, as well as non-genetic medical conditions.

**American College of Medical Genetics and Genomics Screening ACT Sheets** ([www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm))

This is an important resource for abnormal newborn screening results. It provides condition-specific ACT sheets that include a brief description of the condition, what needs to be done after a new diagnosis, and a step-by-step plan-of-action algorithm.

**Genetics in Primary Care Institute** ([www.geneticsinprimarycare.org](http://www.geneticsinprimarycare.org))

This Web site is a clearinghouse of practical tools and information for PCPs. Topics include genetics and genomics, genetic testing, family history, genetic counseling, patient communication, ethical concerns, and more. Brief informational video testimonials from a wide range of professionals provide insight and various points of view on key genetics topics. This Web site also contains an archive of educational webinars hosted by the Genetics in Primary Care Institute, an electronic family history tool, and a quality improvement toolkit, which contains strategies for assessing risk and improving care for patients with genetic-related conditions. The GPCI also developed a PediaLink course, "Dive into the Gene Pool", which will launch in August 2014.