There are many useful Web sites that provide information about initiating a genetic diagnostic work-up and responding to unexpected clinical situations such as an abnormal newborn screening result, prenatal fetal abnormalities or patients who present with multiple congenital anomalies. In addition, there are Web sites that health care providers can refer families to for additional information on genetic conditions, as well as links or contact information for support organizations, according to the webinar presentation, “Top 10 Genetics Resources for Pediatric Primary Care Providers,” held on October 25, 2012. The webinar, which featured Beth A. Pletcher, MD, FAAP, FACMG, was part of the Time Out for Genetics webinar series hosted by the Genetics in Primary Care Institute (GPCI).

What are the Top 10 Genetic Resources?

   - 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.
   - 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.
   - The Web site will reveal multiple conditions that display any or all of the unusual features typed in for the search.
   - Too much non-specific information may not locate the right diagnosis and too little non-specific information may uncover too many conditions.
   - Locating the conditions that list all of the features input for the search will help providers to narrow down the diagnosis.
   - A table of contents includes a synopsis of each genetic condition generated from the search.

   - This Web site is designed for medical professionals.
   - As of February 2013, the Web site includes more than 570 GeneReviews, which are 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.
   - It is important to note that the spelling of a condition must be accurate in order to retrieve information about that condition.
   - 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.
   - The directory of laboratories lists specific testing offered by labs and contact information.

3. **American Academy of Pediatrics (AAP) Committee on Genetics**
   - This resource includes physician-friendly health supervision guidelines for common genetic topics, including achondroplasia, Down syndrome, Fragile X syndrome, Marfan syndrome, neurofibromatosis, Prader-Willi syndrome, sickle cell disease, and Williams syndrome.
   - The health supervision guidelines 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.
Top 10 Genetics Resources for Pediatric Primary Care Providers

   • 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.

5. American College of Medical Genetics and Genomics Screening ACT Sheets [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.

   • This Web site is useful for health care providers as well as families.
   • This well-organized Web site includes a directory of genetic support groups that can be found by clicking on specific genetic disorders, which are listed alphabetically.
   • Links to the support groups, as well as contact information are provided.
   • Parent information on newborn screening tests, similar to the ACMG ACT sheets, can be found at www.babysfirsttest.org.

7. Unique Rare Chromosome Group [http://www.rarechromo.org]
   • This family-friendly Web site includes information only about rare chromosomal disorders.
   • Specific cytogenetic variation leaflets written in English, German, Spanish, French, and Italian can be found on this Web site.

8. Genetics and Rare Conditions [http://www.kumc.edu/gec/support]
   • This Web site has an alphabetic listing of rare conditions and genetic disorders that link to support groups and other Web sites, including those in other countries and in other languages.

   • This Web site for medical providers and families contains descriptions, symptoms, causes and treatments for rare disorders, as well as links to support groups.

10. Genetics in Primary Care Institute [www.geneticsinprimarycare.org]
    • This Web site is a clearinghouse of practical tools and information for primary care providers.
    • Topics include genetics and genomics, genetic testing, family history, genetic counseling, patient communication, 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, as well as a cache of educational resources from external partners, for primary care providers.

About the Presenter
Dr Pletcher is an Associate Professor of Pediatrics at the University of Medicine and Dentistry-New Jersey Medical School in Newark, NJ, and co-director of the Neurofibromatosis Center of New Jersey in Newark. She also serves on the Bioethics Committee of the University Hospital in Newark and is immediate past chair of the American Academy of Pediatrics Committee on Pediatric Workforce.

About GPCI
The GPCI was established to increase primary care providers’ knowledge and skills in the provision of genetic-based services. The GPCI is a cooperative agreement between the US Department of Health and Human Services, the Health Resources & Services Administration, the Maternal & Child Health Bureau and the American Academy of Pediatrics.

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