The Development and Use of the Genetic Testing Electronic Quality Information Portal, GeT-EQuIP

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Background: In 2005 the American College of Medical Genetics (ACMG) recommended a national uniform newborn screening (NBS) panel. Many states have adopted these recommendations, resulting in an increased need by healthcare professionals and the public for information regarding diagnosis, intervention, and management of the disorders. However, the desired information may not always be publicly available. Based on recommendations from the CDC- hosted "Quality, Access, and Sustainability of Biochemical Genetic Testing Working Meeting" in October 2006, we assessed the current information resources for NBS diseases and related genetic testing and explored ways to develop a common information portal for general practitioners, specialists, laboratories, and the general public.

Methods: Twenty commonly used websites were assessed for information regarding basic information, genetic testing information, laboratories, testing algorithm and availability, sensitivity/ specificity of genetic tests, interpretation of test results, and disease management for the 84 diseases on the ACMG NBS panel.

Findings: The quality and quantity of the information varied among the websites. Five websites contained information provided by external links, two contained restricted access, and two contained no applicable information. The most common information elements were basic information and availability of genetic tests for newborn genetic diseases. Many websites lacked information on test algorithms, sensitivity/specificity, interpretation, and disease management.

Conclusions: Availability and accessibility of NBS information is an increasing public health need. Using the information compiled, we have developed a searchable database, GeT-EQuIP, and disease-specific information about NBS conditions. This website is available through the CDC's main website at www.cdc.gov/dls/genetics/GeTEQuIP .