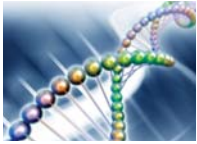


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The genetics encounter: Different from the typical clinic visit

As a practicing pediatric geneticist, I spend significant time with my patients and their families addressing personal and social issues while helping them to cope with the disorders we identify. A visit to a genetics clinic is unique in several ways.

First, our patients are not typically ill when they arrive; they have been referred because of concerns about growth and/or development, unusual physical features, or a family history of a rare disease that have not been resolved by visits to other clinicians. Geneticists perform “detective” work trying to clarify the underlying cause for a patient’s problems.

Second, the patient’s parents often arrive with substantial anxiety, feelings of guilt, and fear about what the future portends. Such personal implications of genetic disorders can be challenging to address as patients and parents learn about a specific disorder and the effects it might have on the family.

Last, while patients often arrive in clinic knowing that they are to see a geneticist, they may have little idea of why they were referred or what will happen in clinic. Sometimes this is a result of having been referred by a health care provider who was unable to articulate exactly why the patient and family need to see a geneticist. Such providers may recognize that a child has a problem, but their lack of familiarity with rare genetic disorders hinders their ability to explain what lies ahead for the child. Frequently, the patients and/

or their parents may be frustrated that an explanation for their medical problems is not readily available. Extensive, time-consuming, and expensive testing, including laboratory studies or diagnostic imaging, may be necessary before an etiology can be identified.

When the patient arrives in clinic, an extensive **medical history** is obtained by a genetic counselor or the physician, and a complete **physical examination** is performed. In addition, the genetic counselor or geneticist will spend significant time obtaining a **family history** to look for clues to a diagnosis. The inheritance pattern of a disorder may become evident as a family pedigree is constructed. If an inheritance pattern is discernible, it can be helpful in limiting the differential diagnosis to only those disorders known to follow this specific pattern.

While the family history is explored and the pedigree is drawn, **social issues** involving the family may become readily apparent. Rarely do we find the “typical” American nuclear family, and the dynamics of a blended family can often be clarified during this part of this visit. Having an understanding of the social situation allows the genetic counselor and clinical geneticist to determine the best way to provide information to the family. The family’s support system and ability to cope with the implications of the disorder can also be assessed.

The geneticist’s approach to the physical examination of a child relies on pattern recognition and the ability to discern clinical features that fall outside the range of normal. The facial gestalt and the general proportions of the patient provide essential clues to a diagnosis. Detailed analysis of numerous **anthropometric assessments** (length/height, weight, head circum-

ference, palpebral fissure length, etc.) may provide additional evidence for a specific diagnosis. Comparison graphs are available for each measurement to determine whether findings fall within the normal range.

While these measurements are helpful, the acumen of the geneticist is crucial to incorporating all the clinical features from the history and physical examination into a unifying diagnosis. Often it is the subtle or minor anomalies that prove most helpful in identifying an underlying etiology. Sometimes the medical geneticist will be aided by **clinical genetics databases** to identify a likely candidate diagnosis.

In addition, the geneticist will also determine if cytogenetic, molecular, biochemical **laboratory studies** and **diagnostic imaging** studies might be helpful. The physician and genetic counselor will explain the rationale for each recommended test and provide the patient and family with guidance on how to obtain coverage for some of the more expensive molecular studies, if they are needed. In an effort to relieve parental concerns about guilt, additional time will be spent providing **education**. For example, the nature of genetic disease may be addressed, and families may be told that although we choose with whom we share our genes, we have no control over how they interact.

The genetics evaluation relies extensively on general medical knowledge, along with a talent for details. With a chronic shortage of clinical geneticists, physician assistants may augment the availability of clinical genetics services while participating in an ever-evolving, exciting, challenging career. [JAAPA](#)

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