

The following list includes some features that might suggest the child has a genetic disorder. However, some of these characteristics are commonly found in people without a disorder:

- Ear abnormalities
- Unusually shaped eyes
- Different colored eyes
- Facial features that are unusual or different from other family members
- Brittle or sparse hair
- Excessive body hair
- White patches of hair
- Large or small tongue
- Misshapen teeth
- Missing or extra teeth
- Loose or stiff joints
- Unusually tall or short stature
- Webbed fingers or toes
- Excessive skin
- Unusual birthmarks
- Increased or decreased sweating
- Unusual body odor
- Abnormal hand creases



## When to Suspect a Genetic Syndrome

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A genetic syndrome is a clinical situation that combines major and minor malformations, anomalies of growth and of psychomotor development.

Although many genetic conditions are individually rare, they are common in aggregate and place a great burden on affected patients and the medical system. Congenital malformations (many of which are related to genetic disturbances) occur in approximately 5 percent of live-born infants; a much higher proportion of conceptions are affected by genetic anomalies. In one study, an estimated 79 of 1,000 live-born infants were found to have a genetic syndrome and/or congenital malformation by 25 years of age.

Prompt recognition of features suggestive of genetic conditions can improve the selection of appropriate, cost-effective diagnostic tests, the performance of well-informed genetic counseling related to issues such as prognosis and future family planning and timely referral to subspecialists.

General themes that can alert pediatricians to the presence of genetic syndromes include dysmorphic features that are evident on physical examination; multiple anomalies in one patient; unexplained neurocognitive impairment; and a family history that is suggestive of a hereditary disease.

Taking an accurate three-generation family history is important when a genetic syndrome is suspected. Important elements include the age and sex of family members; when family members were affected by disease or when they died; the ethnic background; and if there is consanguinity. Every effort should be made to obtain complete information about the gestation leading to the birth of the affected child .

Events surrounding the birth of the child may be critical: antepartum status, length of labor, mode of delivery, baby's condition at birth, measurements. Finally, information is obtained about the child's course since infancy along the lines of a standard pediatric history, but with a few special emphases: general health, growth, developmental progress, behavior, special testing or therapy.

The physical examination is of enormous importance: the basic techniques of physical diagnosis are used, but the examination is fine tuned to promote detection of many subtle physical clues that might otherwise be overlooked. Selected measurements of physical features can be extremely useful in confirming a clinical impression of abnormality. The diagnosis of a particular disorder can be based on clinical features, laboratory data, or a combination of both.