Valley Children's HEALTHCARE

Central California
Pediatrics

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Specialty information for physicians who treat children and expectant mothers.

When to Send a Patient to a Geneticist for Differences in Morphometrics

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Morphometrics: The analysis of form, size and shape variations in children - more commonly referred to as growth parameters.

When growth parameters are abnormal, the next step might be to refer the patient to a geneticist. Prior to this referral, a couple of questions should be considered.

Is the measurement truly out of range? What has the growth trend been over time?

The Centers for Disease Control and Prevention (CDC) and the World Health Organization (WHO) have nomograms depicting normal pediatric growth over time. The third percentile represents two standard deviations below the mean, while the 98th percentile represents two standard deviations (SD) above the mean. Patients whose measures fall outside of two standard deviations may still be normal if the measurements are consistent within the context of the family. For example, a child whose height is at the third percentile would not be inconsistent if parental heights were in similar range.

Even when measurements do seem consistent with familial expression, those falling significantly above or below two standard deviations can still represent the expression of a syndrome. Therefore, of equal importance is the trend of growth (or lack thereof) over time, and whether or not the measurement has shifted percentiles. It is normal for children to follow consistently along a "channel" or between the same two percentile lines over time. The unexpected crossing of two or more percentile lines should give pause and raise the question of whether or not a genetic syndrome or occult systemic illness is present.

What if more than one measurement falls outside of two standard deviations?

While many syndromes may affect one or more growth parameters, systemic illness should also be considered. The combination of low height and weight is often the result of a problem with a specific body system such as gastrointestinal, endocrine, pulmonary, renal or cardiac. If signs or symptoms of systemic illness are present, e.g. inflammatory bowel disease or celiac disease, these should be ruled out first. In contrast, if one of the abnormal growth parameters is head circumference, this warrants a more urgent genetic consultation – especially in the context of developmental delay or intellectual disability.

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Outside of growth parameters, what else on physical exam may raise the index of suspicion?

While major anomalies, such as limb reduction defects, may clearly point to a possible underlying genetic condition, minor anomalies can be much more subtle. Minor anomalies are phenotypic features that do not compromise normal bodily function. These can include, but are not limited to, multiple hair whorls, epicanthal folds, ear tags, single palmar crease, clinodactyly, unusual ear folds, short or abnormally slanted palpebral fissures, sandal gap deformity and pectus excavatum. Most of the typical population inherits one or two minor anomalies from a parent. The presence of three or more minor anomalies should prompt a referral for genetic consultation

How important is family history?

Family history is key in identifying genetic conditions, and can provide valuable information with respect to patterns of inheritance. Is there a family history of neurodevelopmental delay or regression? Have there been problems with pregnancy, infertility or birth defects in the family? Are there genetic conditions or syndromes associated with the patient's ethnicity? Have family members died or become sick at an early age? Do multiple family members share a constellation of conditions? Is there consanguinity? A positive answer may be a clue into an underlying genetic diagnosis.

How can providers help get the most out of a Genetics referral for morphometric abnormalities?

Complex traits like height and weight are influenced by a staggering number of genes working in tandem within the context of the patient's environment. The genetic work up can be optimized by providing our team with a copy of the child's growth curves, any physical exam findings and laboratory/radiologic results previously obtained.

Our medical genetics and metabolism team is the largest in the Central Valley and is committed to using genetic testing and risk assessment to optimize the long-term health of children. In a capacity that is ever changing, we can work together to help families.

The Valley Children's Department of Genetics remains available always to help navigate the questions above.
To contact Valley Children's genetics team, call 559-353-6400.

Upcoming CME Opportunities

Pediatric Clinical Symposium: Top 10 Genetic Referrals – When and When Not To

Presented by Dr. Richard Sidlow
Tuesday, May 26
12:15 p.m. - 1:15 p.m.

Hear Dr. Sidlow expand on differences in morphometrics and other considerations when referring to a geneticist.

Register for this CME at cmetracker.net/VCH

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