Polydactyly

Posted 3-29-05

Key Points

- The term polydactyly refers to the presence of an additional digit (finger or toe). Polydactyly can be postaxial (on the ulnar or fibular side of the limb), preaxial (on the radial or tibial side of the limb) or medial (in the middle of the hand or foot), and can be unilateral or bilateral. The extra digit may be fully or partially developed, and with or without bone.
- Polydactyly can occur as an isolated malformation or as part of a genetic syndrome.
- Isolated (nonsyndromic) postaxial polydactyly is relatively common and may be inherited in an autosomal dominant manner with reduced penetrance.
- Family communication about a history of isolated postaxial polydactyly is encouraged to prevent unnecessary medical evaluations by physicians and to minimize parental concern during pregnancy or in the newborn period.

Learning Objectives

Participants will be able to:

- Calculate the risk of a child having isolated postaxial polydactyly in a family demonstrating autosomal dominant inheritance with incomplete penetrance;
- Explain the need for family communication regarding genetic conditions to help prevent unnecessary medical evaluations and minimize parental concern.

Family History Issues

Isolated postaxial polydactyly is usually caused by a gene mutation which is inherited in an autosomal dominant manner with incomplete penetrance, meaning that individuals who have a parent with isolated postaxial polydactyly have a 50% chance of inheriting the gene mutation but may not manifest the polydactyly. As a result, it may appear to "skip" a generation.

Since polydactyly is usually repaired early in life and may be forgotten or not discussed in families, ascertaining a complete family history of polydactyly may be difficult.

Red Flags 

About 15% of children born with polydactyly have other congenital anomalies, usually as part of a defined syndrome. This is true for about 10% of children with postaxial polydactyly, 20% of those with preaxial polydactyly, and more than 50% of those with rare polydactylies [Castilla et al 1998]. Unless there is a clear family history of isolated polydactyly, any newborn with polydactyly should be investigated for the presence of associated anomalies. Hand postaxial polydactyly and foot postaxial polydactyly are considered different traits. Hand postaxial polydactyly is more frequent (75%) than foot postaxial polydactyly (15%), and is less often found with other anomalies (7% vs 15%). Having both hand and foot postaxial polydactyly is the least frequent occurrence (10%) and is associated with the highest frequency of associated anomalies (23%) [Castilla et al 1997].

Case 31. Family History of Extra Finger (Polydactyly)

Mr. G was born with six fingers on each hand. Based on what he was told as a child, he describes the extra fingers as "extra fifth fingers." They were removed when he was an infant. He is now 35 years old, works as a mechanical engineer, and has no health problems. He is planning to get married soon. Both his parents have normal hands, but a paternal uncle and his paternal grandmother were also both born with extra fifth fingers. His mother told him that extra fingers are "genetic" and that his children would have them too. However, his aunt said not to worry because the problem typically skips generations. He wants to check this out before discussing it with his fiancee.

Clinical Care Issues

Isolated versus syndromic polydactyly

Postaxial polydactyly — the presence of a fully formed or rudimentary extra digit next to the little finger or little toe — typically occurs as an isolated malformation, without other health consequences. From the history Mr. G provides, this is likely to be his diagnosis; further confirmation can be derived from examining his hands for surgical scars and by obtaining a description of his hands before surgery from his birth records or medical chart. Some inherited syndromes, such as Meckel-Gruber syndrome, Ellis van Creveld syndrome, short rib-polydactyly syndrome, Smith-Lemli-Opitz syndrome, trisomy 13, and Bardet-Biedl syndrome, can include polydactyly, but usually also include other obvious congenital anomalies [Jones 1997, Castilla et al 1998, Krakow 2002]. Mr. G doesn't seem to have any associated anomalies, and a syndromic cause is unlikely in his case. If a syndrome were diagnosed, the children of an affected person might be at risk for additional health problems associated with that particular syndrome, if it is inherited in an autosomal dominant manner; or the children might not be at risk at all, if the disorder is inherited in an autosomal recessive manner and if there is no reason to suspect that his fiancee could be a carrier of the disorder.

Isolated hand postaxial polydactyly is much more common in African Americans than in other ethnic groups. If found in a newborn of another ethnic group, an underlying syndromic cause should be suspected. On the other hand, isolated hand preaxial polydactyly is most commonly found in whites. It is most often unilateral and sporadic [Kozin 2003].

Risk of polydactyly in Mr. G's children

Family history is useful in evaluating polydactyly because isolated postaxial polydactyly is usually inherited in an autosomal dominant manner. The family history helps to confirm both the expected inheritance pattern and the lack of other medical complications in family members with polydactyly. In Mr. G's case, his father was not affected, but his father's mother and father's brother were affected. This history is consistent with autosomal dominant inheritance with incomplete penetrance — that is, a condition in which some people with the mutation do not have clinical manifestations of the disorder. (Incomplete penetrance often results in the perception that disorders "skip generations.")

Risk Assessment

Assuming the diagnosis is autosomal dominant isolated postaxial

polydactyly, Mr.G has a 50% chance of passing on the gene mutation to each child. Because of incomplete penetrance, a child who inherits the mutation will not necessarily manifest polydactyly. Thus, the likelihood that a particular child will have polydactyly is somewhat lower than 50%. However, penetrance appears to be relatively high in autosomal dominant isolated postaxial polydactyly, so the likelihood that a particular child will have polydactyly remains significant.

Genetic Counseling and Testing

Although more than one gene associated with isolated postaxial polydactyly has been identified, genetic testing is not clinically available. If available, genetic testing could confirm clinical diagnosis, but would be unlikely to change clinical management of the patient or his offspring. If Mr. G is concerned that a fetus may have polydactyly, prenatal ultrasound examination could be offered during a pregnancy to determine whether a sixth digit is visible. Extra digits should be visible as early as 13 weeks of gestation. It is important to explain to Mr. G that the ability to visualize an extra digit on ultrasound examination depends upon the time during gestation when the ultrasound examination is done, the experience of the operator, the activity/position of the fetus, and the specific configuration and location of the extra digit. Ultrasound can also be used to look for associated anomalies [Bromley et al 2000, Zimmer & Bronshtein 2000].

Interventions

The options for treatment of isolated polydactyly depend on the characteristics of the extra digit. If it is rudimentary and pedunculated (type B), its base can be tied with a suture in the newborn period, and it will fall off spontaneously. This procedure can sometimes leave a small bump at the base. Better developed extra digits (type A), especially those containing bone or cartilage, can be corrected with surgery. This procedure is usually done during the first year of life [Kozin 2003].

Ethical/Legal/Social/Cultural Issues

Family communication

Mr. G should be encouraged to share his family history of polydactyly with his future wife so that she can report this to her obstetrician. If polydactyly were incidentally identified on a fetal ultrasound examination or in the newborn period without this history available, medical evaluations might be

undertaken for the many genetic syndromes that include polydactyly.

Additionally, if his wife knows about Mr. G's family history of polydactyly, she will be aware that there is a chance that her child may have polydactyly, and will be prepared if their child is born with extra digits.

Concept of normal

The importance of appearance and body image varies among cultures. Today in Western society, having a "normal" appearance is important; however, the concept of normal is difficult to define. What makes a person's body abnormal to the point that medical treatment is necessary? This question can be asked for a wide range of physical traits, including obesity, large breasts, and birth marks. When treatment is offered to change or "correct" appearance, the distinction between cosmetic and indicated medical treatment can sometimes be difficult to define. Polydactyly can be corrected through a simple surgical procedure, but is the "abnormality" sufficient to put a child through surgery? Although surgery is routinely offered for polydactyly, parents may differ in their opinion about the need for surgical correction. Even as medical technology advances, physicians need to respect the choice of parents and individuals in favor of the "non-normal."

However, some types of extra digits may be removed for reasons other than appearance. Postaxial rudimentary pedunculated extra digits (type B) are not functional: the child has no control over mobility of the digit, and it can get caught in clothing, etc. There is a strong possibility that it will get "torn off"; thus, removal in the newborn period is preferred. Extra digits might hinder normal hand function, especially if they are combined with other finger anomalies (e.g., syndactyly, camptodactyly). In these cases, surgery is indicated to restore normal hand function.

Resources

- **Online Mendelian Inheritance in Man: Postaxial polydactyly type A1**
- **General Practice Notebook - a UK medical encyclopedia** *Site includes:*

- Some general information on polydactyly, cleft hand, syndactyly, camptodactyly, and clinodactyly
- Diseases associated with polydactyly and syndactyly

- **Emedicine: Supernumerary Digit** *Site includes:*
  - Basic background and pathophysiology of polydactyly
  - Differential diagnosis and syndromes associated with polydactyly

- **Emedicine: Polydactyly of the Foot** *Site includes:*
  - An overview of the surgical correction of polydactyly
  - Complications, outcomes and prognosis
  - Multiple pictures of polydactyly of the foot

- **PennState: Milton S Hershey Medical Center, College of Medicine**

- **Birth Disorder Information Directory** *Site includes:*
  - Information about both polydactyly and syndactyly
  - Links to OMIM for different types of each disease
  - Pictures
- **GeneTests Online Medical Genetics Information Resource**

References

Bromley B, Shipp TD, Benacerraf B (2000) Isolated polydactyly: prenatal diagnosis and perinatal outcome. *Prenat Diagn* 20:905-8 [Medline]

Castilla EE, Lugarihno R, da Graca Dutra M, Salgado LJ (1998) Associated anomalies in individuals with polydactyly. *Am J Med Genet* 80:459-65 [Medline]

Castilla EE, Lugarihno da Fonseca R, da Graca Dutra M, Paz JE (1997) Hand and foot postaxial polydactyly: two different traits. *Am J Med Genet* 73:48-54 [Medline]

Jones KL (1997) Smith's Recognizable Patterns of Human Malformation, 5th ed. WB Saunders, Philadelphia. p 816

Kozin SH (2003) Upper-extremity congenital anomalies. *J Bone Joint Surg Am* 85:1564-76 [Medline]

Krakow D (2002) The dysostoses. In: Rimoin DL, Connor JM, Pyeritz RE, Korf BR (eds) Emery and Rimoin's Principles and Practice of Medical Genetics, 4th ed. Churchill

Livingstone, New York. pp 4160-81

Zimmer EZ and Bronshtein M (2000) Fetal polydactyly diagnosis during early pregnancy. *Am J Obstet Gynecol* 183:755-8 [Medline]