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REVIEW ARTICLE

POLYDACTYLY – A REVIEW

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ABSTRACT

Polydactyly is a condition in which a person has more than five fingers per hand or five toes per foot. Having an abnormal number of digits (6 or more) can occur on its own, without any other symptoms or disease. Polydactyly may be passed down (inherited) in families. This trait involves only one gene that can cause several variations. African Americans, more than other ethnic groups, can inherit a 6th finger. In most cases, this is not caused by a genetic disease. Polydactyly can also occur with some genetic diseases. Extra digits may be poorly developed and attached by a small stalk (generally on the little finger side of the hand). Or, they may be well-formed and may even function. Poorly formed digits are usually removed. Simply tying a tight string around the stalk can cause it to fall off in time if there are no bones in the digit. Larger digits may need surgery to be removed. The doctor should ask the parents whether there was polydactyly at birth, because a person may not know they have it.

INTRODUCTION

Polydactyly or polydactylism (from Greek origin, *polys* meaning "many", and *daktylos* meaning "finger" also known as hyperdactyly, is a congenital physical anomaly in humans, dogs, and cats having supernumerary fingers or toes. Polydactyly is the opposite of oligodactyly which indicates fewer fingers or toes. The extra digit is usually a small piece of soft tissue that can be removed.[1] Occasionally it contains bone without joints; rarely it may be a complete, functioning digit. The extra digit is most common on the ulnar (little finger) side of the hand, less common on the radial (thumb) side, and very rarely within the middle three digits. These are respectively known as postaxial (little finger), preaxial (thumb), and central (ring, middle, index fingers) polydactyly. The extra digit is most commonly an abnormal fork in an existing digit, or it may rarely originate at the wrist as a normal digit does. Polydactyly belongs to the category of duplication. The extra fingers are usually smaller than his other fingers and are abnormally developed. The extra fingers or toes may be small and non-functional, in the case of nubbins or skin tags, or they may be fully formed with bones and skeletal connections. Polydactyly often occurs bilaterally (on both hands or both feet), or it may occur on just one hand or foot. Similarly, a person may have extra digits on just the hands, just the feet, or some other combination.[2][3]

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Causes of Polydactyly

Polydactyl occurs when the body follows a different set of directions than usual while forming the hands or feet during development. Researchers are still learning about all the genes that cause extra digits. The trait may be passed down in families as an isolated, benign condition, like having a hitchhiker's thumb or being double jointed. This is considered a non-syndromic anomaly.[4] Alternatively, the trait may exist as part of a syndrome, which is a group of several recognizable clinical features that often occur together. Some syndromes that might present with polydactyly include Greig Cephalopolysyndactyly Syndrome (GCPS) or Bardet-Biedl Syndrome (BBS). The causes are[5] :

- Asphyxiating thoracic dystrophy
- Carpenter syndrome
- Ellis-van Creveld syndrome (chondroectodermal dysplasia)
- Familial polydactyly
- Laurence-Moon-Biedl syndrome
- Rubinstein-Taybi syndrome
- Smith-Lemli-Opitz syndrome
- Trisomy 13

Genetic Causes

Polydactyly is associated with different mutations, either mutations in a gene itself or in a cis-regulatory

element responsible for the expression of a specific gene. In many cases Sonic hedgehog (Shh) is affected, rarely Indian Hedgehog (double foot mutant) or Bone morphogenetic proteins (BMP).[6] Also mutations in Hoxa- or Hoxd clusters are reported leading to polydactyly. Interactions of Hoxd13 and GLI3 induce synpolydactyly, a combination of extra and consolidated digits. Other signal transduction pathways in this context are the Wnt signaling pathway or Notch.In a concrete case of preaxial polydactyly (Hemingway mutant) a mutation of the cis-regulatory element ZRS (ZPA regulator sequence) is associated. ZRS is a noncoding element, 800.000 basepairs remote to the target gene Shh. An ectopicexpression of Shh is seen on the anterior side of the limb. Normally Shh is expressed in an organiser region, called the zone of polarizing activity (ZPA) on the posterior limb side. From there it diffuses anteriorly, laterally to the growth direction of the limb. In the mutant mirroring smaller ectopic expression in a new organiser region is seen on the posterior side of the limb. This ectopic expression causes cell proliferation delivering the raw material for one or more new digits.[7]

Congenital Polydactyly

Polydactyly occurs in the womb as a detour on the road to developing just one thumb and four fingers on the hand. In the womb, the new hand starts out in the shape of a paddle, then splits into separate fingers. Sometimes the fingers don't split apart enough, and webbed fingers result: syndactyly. Sometimes a extra split forms and extra fingers result: polydactyly. Syndactyly and polydactyly are about equally common disorders. Combinations of both can occur as well - webbed extra fingers. Sometimes these problems are in the genes and can be passed down generation to generation, but many times there is simply no known explanation. [8]

Symptoms of Polydactyly

Children with polydactyly are born with one or more extra fingers or toes.

The extra digit may be:

- A small, raised bump or a piece of skin that looks like a small finger or toe that isn't fully formed (nubbin)
- A complete, working finger or toe
- For some children, polydactyly is only one feature of a more complex genetic condition or syndrome. These children will have other signs and symptoms.[9]

Inheritance of Polydactyly

African Americans, more than other ethnic groups, can inherit a 6th finger. In most cases, this is not caused by a genetic disease.Polydactyly can also occur with some genetic diseases.The condition has an incidence of 1 in every 500 live births. Postaxial hand polydactyly is a common isolated disorder in African black children, and autosomal dominant transmission is suspected. Postaxial polydactyly is more frequent in blacks than in whites and is more frequent in male children. In contrast, postaxial polydactyly seen in white children is usually syndromic and associated with an autosomal recessive transmission. One study by Finley et al. combined data from Jefferson County, Alabama, United States and Uppsala County, Sweden. This study showed incidence of all types of polydactyly to be 2.3 per 1000 in white males, 0.6 per 1000 in white females, 13.5 per 1000 in black males, and 11.1 per 1000 in black females. African Americans, more than other ethnic groups, can inherit a 6th finger.[10] In most cases, this is not caused by a genetic disease.Polydactyly can also occur with some genetic diseases. Research has shown that the majority of congenital anomalies occur during the 4-week embryologic period of rapid limb development. Polydactyly has been associated with 39 genetic mutations. More specific loci and genetic mechanisms responsible for disorders of duplications will be defined with time, as molecular research continues.[11]

Forms of Polydactyly

There are many different forms of polydactyly,[12] ranging from

- a small extra bump on the side of the hand,
- a finger which widens to end in two fingertips,
- an extra finger which dangles by a thin cord from the hand,
- a hand which looks normal except that it has a thumb and five fingers, and
- an infinite number of other variations.

Types of Polydactyly

Ulnar or postaxial polydactyly: This is the most common situation, in which the extra digit is on the ulnar side of the hand, thus the side of the little finger. This can also be called postaxial polydactyly. It can manifest itself very subtly, for instance only as a nubbin on the ulnar side of the little finger, or very distinctly, as a fully developed finger. Most commonly, the extra finger is rudimentary, consisting of an end phalanx with a nail, and connected to the hand with a small skin pedicle. Mostly one neurovascular bundle can be identified, with no tendons present in the extra digit. In case of a fully developed extra finger, the duplication usually presents itself at the level of the metacarpophalangeal joint.

A triplication of the little finger is very rare. Ulnar polydactyly occurs ten times more often in negroid ethnicities and is most common in African populations. The incidence in Caucasians is reported as 1 in 1,339 live births, compared with 1 in 143 live births in Africans and African Americans.[13][14] Ulnar polydactyly is also often part of a syndrome. In patients with African ancestry ulnar polydactyly mostly occurs isolated, whereas the presentation in Caucasians is often associated with a syndrome. In almost 14% of all patients, this type of polydactyly is hereditary. It usually passes on in an autosomal dominant manner with variable expression and incomplete penetrance. It is genetically heterogenic, meaning that mutations in different genes can be the cause. When the extra digit is pedicled it can be very movable; however, injuries are rare and have never been reported so far. The classification of ulnar polydactyly exists of either two or three types. The two-stage classification according to Temtamy and McKusick involves type A and B. In type A there is an extra little finger at the metacarpophalangeal Joint, or more proximal including the Carpometacarpal joint. The little finger can be hypoplastic or fully developed. Type B varies from a nubbin to an extra,

non-functional little finger part on a pedicle.Sometimes that can be from a radiation. According to the three-type classification, type I includes nubbins or floating little fingers, type II includes duplications at the MCPJ, and type III includes duplications of the entire ray.[15][16][17]

Radial or preaxial polydactyly

This is a less common situation, which affects the side of the hand towards the thumb. Radial polydactyly refers to the presence of an extra digit (or extra digits) on the radial side of the hand. It is most frequent in Asian populations and it is the second most common congenital hand disorder. The incidence of radial polydactyly is reported as 1 in every 3,000 live births. The clinical features of radial polydactyly will depend upon the extent of duplication. Radial polydactyly varies from a barely visible radial skin tag to complete duplication. Thumb polydactyly varies from barely visible broadening of the distal phalanx to full duplication of the thumb including the first metacarpal. Radial polydactyly is frequently associated with several syndromes. The Wassel classification is the most widely used classification of radial polydactyly. It is based upon the most proximal level of skeletal duplication:[18]

- Type 1: distal phalanx
- Type 2: interphalangeal joint
- Type 3: proximal phalanx
- Type 4: metacarpophanlangeal joint
- Type 5: metacarpal phalanx
- Type 6: carpometacarpal joint
- Type 7: triphalangeal thumb

The most common type is Wassel 4 (about 50% of such duplications) followed by Wassel 2 (20%) and Wassel 6 (12%).[19]

Central Polydactyly

This is a very rare situation, in which the extra digit is on the ring, middle or index finger. Of these fingers, the index finger is most often affected, whereas the ring finger is rarely affected. This type of polydactyly can be associated with syndactyly, cleft hand and several syndromes. Polysyndactyly presents various degrees of syndactyly affecting fingers three and four. The classification of central polydactyly is based on the extent of duplication and involves the following three types: Type I: a central duplication, not attached to the adjacent finger by osseous or ligamentous attachments. It frequently does not consist of bones, joints, cartilage, or tendons. Type IIA: a nonsyndactylous duplication of a digit or part of a digit with normal components and articulates with a broad or bifid metacarpal or phalanx. Type IIB: a syndactylous duplication of a digit or part of a digit with normal components and articulates with a broad or bifid metacarpal or phalanx. Type III: a complete digital duplication, which has a well-formed duplicated metacarpal. [20][21]

Diagnosis

Tests used to diagnose the condition:

- Chromosome studies
- Enzyme tests
- X-rays
- Metabolic studies

During pregnancy, this condition may be diagnosed with ultrasound or a more advanced test called embryofetoscopy during the first three months.Your child may need an X-ray to see if there are bones in the extra digit and to see how the extra digit connects to the rest of the hand or foot. If the doctor thinks your child might have other genetic problems, your child might have tests to check their chromosomes. [22][23]

Syndromes Associated with Polydactyly

Ulnar polydactyly is often bilateral and associated with syndactyly and polydactyly of the feet. This can be a simple or complex polydactyly. Ulnar polydactyly occurs as an isolated congenital condition, but can also be part of a syndrome. The syndromes which occur with ulnar polydactyly are: Greig cephalopolysyndactyly syndrome, Meckel syndrome, Ellis–van Creveld Syndrome, McKusick–Kaufman syndrome, Down syndrome, Bardet–Biedl syndrome, Smith–Lemli–Opitz syndrome.[24] Type VII of radial polydactyly is associated with several syndromes: Holt–Oram syndrome, Fanconi anemia: aplastic anemia at the age of 6, Townes–Brocks syndrome, Greig cephalopolysyndactyly: also occurs with ulnar polydactyly. The syndromes associated with central polydactyly are: Bardet–Biedl syndrome, Meckel syndrome, Pallister–Hall syndrome, Legius syndrome, Holt–Oram syndrome, Also, central polydactyly can be associated with syndactyly and cleft hand. Other syndromes including polydactyly include Acrocallosal syndrome, Basal cell nevus syndrome, Biemond syndrome, Ectrodactyly-ectodermal dysplasias-cleft lip/palate syndrome, Mirror hand deformity, Mohr syndrome, Oral-facial-digital syndrome, Rubinstein-Taybi syndrome, Short rib polydactyly, and VATER association.[25][26] It can also occur with a triphalangeal thumb.

Treatment of polydactyly

In most cases, doctors remove an extra finger or toe in early childhood. The goal of treatment is to give your child a hand or foot that works well and looks typical. The method for removing an extra digit depends on how it connects to the hand or foot. An extra digit may connect with only a narrow stalk of tissue, or it may connect more deeply and share bones, muscles and other tissues with the hand or foot.

Vascular clip: If the digit is poorly formed and contains no bone, sometimes the treatment is as simple as attaching a vascular clip at the base during a clinic visit. The clip stops blood flow to the digit so it will fall off, like the stump of belly button does soon after birth. After attaching the clip, the doctor puts a bandage on your child's hand or foot. In a couple of weeks, your child comes back to the clinic to have the bandage removed.[27]

Surgery: If the digit is better formed, a surgeon removes it in the operating room when your child is about 1 year old. This is done as a day surgery. Your child's surgery will be based on their exact condition. More complex cases may require complex surgery. The surgery may involve carefully cutting through or around bones, ligaments, muscles,tendons and other

tissues to remove the extra digit. Then the surgeon may need to move or reconnect some structures before closing the skin so the whole hand or foot works well and looks normal.After surgery, your child may need to wear a cast or splint on their hand or foot while it heals. The doctor will want your child to come back for follow-up visits to make sure they are healing well. Some children who have extensive surgery with cutting through many tissues may have occupational therapy to help with swelling, scarring and stiffness.[28][29]

Procedure of Treatment

The reconstruction does not simply involve an ablation of the smaller duplication portion. The shared articular surface must be reshaped, the bones realigned, and the collateral ligaments of the joint must be reconstructed to ensure the stability of that joint, and tendons must be realigned. If these general principles are not adhered to, the thumb will otherwise have an unstable metacarpal phalangeal joint, and significant residual deformity, and major functional problems.[30] On occasion, because of joint surface malalignment (particularly when the duplication involves a shared joint in addition to the ligament and tendon readjustments) a corrective bony osteotomy may be considered. This procedure involves actually tacking the bone and resetting its angle and may be required at the initial correction of the polydactyly. This is done to avoid any long-term misalignment of the reconstructed thumb and to minimize the risk of a residual zigzag unstable deformity to that reconstruction. [31] While it is important to meet with the child at a young age, within a month or two of birth, the actual reconstructive surgery for polydactyly (digital duplication) will generally be performed somewhere around 9 or 10 months of age. [32]

Conclusion

Polydactyly does not generally pose any health risk over time. Surgical treatment is more likely to result in a satisfactory outcome if surgery is done in the first few years of life to give the child the most ability to adapt and accommodate to the changes in their hand. People are less likely to be satisfied with surgery for polydactyly if surgery is delayed past early childhood.A hand therapist can help with some problems before and after surgery, tailored to the individual problem and the temperament of the child, but some children proceed through their surgery and recovery without needing therapy. The goals of surgery are to improve the appearance of the hand and to prevent progressive deformity from developing as the child grows. Surgery is generally successful in both of these areas, largely correcting the appearance and social stigmata of congenitalism.In many cases, surgery results in a greatly improved but not perfectly normal appearance, and in some situations a normal appearance can not be expected. For example, each of the duplicated thumbs in the above diagram is likely to be smaller than a normal thumb, and no amount of surgery will allow a smaller thumb to "catch up" and perfectly match a normal size thumb on the opposite hand.Fortunately, unless there is something particularly eye-catching about the hand (like an extra finger or thumb), what people notice about another person's hand is not the appearance of an individual finger or thumb, but how the person uses their hand. Hiding one's hand actually draws attention to it, and if surgery allows the person to use their hand in a natural, unselfconscious way, many small details, such as a somewhat small thumb, will go unnoticed.

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