GENEReviews

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Pallister-Hall Syndrome

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Summary

Disease characteristics. Pallister-Hall syndrome (referred to as PHS in this entry) is characterized by a spectrum of anomalies ranging from polydactyly, asymptomatic bifid epiglottis, and hypothalamic hamartoma at the mild end to laryngotracheal cleft with neonatal lethality at the severe end. Individuals with mild PHS may be incorrectly diagnosed as having isolated postaxial polydactyly type A. Individuals with PHS can have pituitary insufficiency and may die as neonates from undiagnosed and untreated adrenal insufficiency.

Diagnosis/testing. The diagnosis of Pallister-Hall syndrome is based on family history and the clinical findings of hypothalamic hamartoma, central and postaxial polydactyly, bifid epiglottis, imperforate anus, and renal abnormalities. Molecular genetic testing of *GLI3*, the only gene known to be associated with Pallister-Hall syndrome, is available clinically.

Management. *Treatment of manifestations:* urgent treatment for endocrine abnormalities, especially cortisol deficiency; management of epiglottic abnormalities depending on the abnormality and the extent of respiratory compromise; bifid epiglottis, the most common abnormality, typically does not need treatment. Standard treatment of anal atresia or stenosis; symptomatic treatment of seizures; elective repair of polydactyly; developmental intervention or special education for developmental delays. *Prevention of secondary complications:* Biopsy or resection of hypothalamic hamartoma may result in complications and lifelong need for hormone replacement; seizures may begin or worsen with use of stimulants for attention deficit disorder. *Surveillance:* during childhood, annual developmental assessment and annual medical evaluation to assess growth and monitor for signs of precocious puberty.

Genetic counseling. Pallister-Hall syndrome is inherited in an autosomal dominant manner. Individuals with PHS may have an affected parent or may have the disorder as the result of a *de novo* mutation. The proportion of cases caused by *de novo* mutations is unknown, as the frequency of subtle signs of the disorder in parents has not been thoroughly evaluated and molecular genetic data are insufficient. The risk to offspring of an affected individual is 50%. Prenatal testing for pregnancies at increased risk is possible if the disease-causing mutation in the family is known. The reliability of ultrasound examination for prenatal diagnosis is unknown.

Diagnosis

Pallister-Hall syndrome (PHS) can be diagnosed based on clinical findings in individuals with classic signs. Molecular genetic testing may be useful to confirm the diagnosis in these

individuals and is used to establish the diagnosis in individuals in whom the clinical findings are ambiguous or mild.

Clinical Diagnosis

Major findings are the following:

- **Hypothalamic hamartoma,** a non-enhancing mass in the floor of the third ventricle posterior to the optic chiasm that is isointense to grey matter on T1 and T2 pulse sequences of an MRI, but may have distinct intensity on FLAIR (Neither cranial CT examination nor cranial ultrasound examination is adequate for diagnosis of hypothalamic hamartoma.)
- **Central (i.e., insertional or mesoaxial) polydactyly,** the presence of six or more well-formed digits with a 'Y'-shaped metacarpal or metatarsal bone
- **Postaxial polydactyly (PAP) types A and B.** PAP-A is the presence of a well-formed digit on the ulnar or fibular aspect of the limb. PAP-B is the presence of a rudimentary digit or nubbin in the same location. Postaxial polydactyly is probably more common than central polydactyly; however, the nonspecificity of postaxial polydactyly and the high frequency of postaxial polydactyly type B in persons of central African descent require caution in its use as a diagnostic feature.
- **Bifid epiglottis,** a midline anterior-posterior cleft of the epiglottis that involves at least two-thirds of the epiglottic leaf. It is a useful feature for clinical diagnosis because it appears to be very rare in syndromes other than PHS and is also rare as an isolated malformation.
- **Other.** Imperforate anus, renal abnormalities including cystic malformations, renal hypoplasia, ectopic ureteral implantation, genitourinary anomalies including hydrometrocolpos, pulmonary segmentation anomalies including bilateral bilobed lungs, and non-polydactyly skeletal anomalies including short limbs

The diagnosis is established in the following individuals:

- A proband if central polydactyly and hypothalamic hamartoma are present
- The first-degree relative of a proband if hypothalamic hamartoma or central or postaxial polydactyly are present (Postaxial polydactyly type B can be used as a diagnostic criterion for first-degree relatives only in persons who are not of central African descent.)

Individuals with postaxial (but not central) polydactyly and a hypothalamic hamartoma **or** central polydactyly without hypothalamic hamartoma **or** hypothalamic hamartoma and other non-polydactyly malformations should be considered for *GLI3* sequencing.

Note: The phenotyping issues for persons with atypical phenotypes and *GLI3* mutations are complex and beyond the scope of this review.

Molecular Genetic Testing

GeneReviews designates a molecular genetic test as clinically available only if the test is listed in the GeneTests Laboratory Directory *by either a US CLIA-licensed laboratory or a non-US clinical laboratory. GeneTests does not verify laboratory-submitted information or warrant any aspect of a laboratory's licensure or performance. Clinicians must communicate directly with the laboratories to verify information.*—ED.

Gene. *GLI3* is the only gene known to be associated with Pallister-Hall syndrome.

Clinical testing

- **Targeted mutation analysis.** Testing for the *GLI3* mutations c.2009delG (p.Gly670GlufsX21) and c.2020delG (p.Glu674SerfsX17) is available on a clinical basis. The utility of testing for these two mutations is unknown (see Table 3; pdf).
- **Sequence analysis.** Sequence analysis of the *GLI3* gene is available clinically. Unpublished data suggest that approximately 95% of typically affected individuals have mutations in the coding region of *GLI3*.

Table 1 summarizes molecular genetic testing for this disorder.

Table 1. Molecular Genetic Testing Used in Pallister-Hall Syndrome

Test Method	Mutations Detected	Mutation Detection Frequency by Test Method	Test Availability
Targeted mutation analysis	c.2009delG c.2020delG [1, 2]	Unknown	Clinical Testing
Sequence analysis	*GLI3* mutations	~95%	

1. The utility of testing for these two mutations is unknown.
2. See Table 3 (pdf).

Interpretation of test results. For issues to consider in interpretation of sequence analysis results, click here.

Testing Strategy

Confirmation of the diagnosis in a proband. Molecular genetic testing is used when the clinical findings are ambiguous or mild.

Prenatal diagnosis for at-risk pregnancies requires prior identification of the disease-causing mutation in the family.

Genetically Related (Allelic) Disorders

Other phenotypes associated with mutations in *GLI3*:

- **Greig cephalopolysyndactyly syndrome (GCPS)** includes polydactyly that is commonly preaxial and may also be postaxial. The polydactyly is commonly associated with cutaneous syndactyly. GCPS has craniofacial features that include ocular hypertelorism, broad forehead, and macrocephaly. Central polydactyly and osseous syndactyly of the metacarpals are not part of GCPS. Most individuals with GCPS have mutations that cause haploinsufficiency of *GLI3*, although a few individuals with a point mutation have been described. However, it has not been demonstrated that these point mutations have stable mRNA or protein. If the stability of these molecules were reduced, the point mutations would result in functional haploinsufficiency. At least one individual with preaxial polydactyly type IV (PPDIV) has been reported to have a *GLI3* mutation [Radhakrishna et al 1999], a finding consistent with clinical suspicion that PPDIV is a mild form of GCPS, in which the limb findings occur without the craniofacial features. However, the craniofacial findings are subtle and there is controversy regarding the distinction of nonsyndromic PPDIV from mild GCPS [Biesecker 2006].
- **Isolated postaxial polydactyly type A (PAP-A).** Mutations in *GLI3* have been identified in individuals with PAP-A [Radhakrishna et al 1999]. However, the PAP-A phenotype has also been shown to result from mutations in other genes.
- **Isolated preaxial polydactyly type IV (PPDIV)** comprises preaxial polydactyly of the hands and/or feet in the absence of other malformations. The severity of the PPDIV

is highly variable [Everman 2005]. Because macrocephaly occurs in the general population and is common in GCPS, the presence of macrocephaly in a person with apparently isolated PPDIV is difficult to interpret.

Clinical Description

Natural History

Pallister-Hall syndrome (PHS) displays a wide range of severity. The literature frequently reflects the assumption that PHS is severe and Greig cephalopolysyndactyly syndrome is mild. This is clearly incorrect, as a minority of individuals with PHS show multiple severe anomalies and most individuals with PHS are mildly affected with polydactyly, asymptomatic bifid epiglottis, and hypothalamic hamartoma. Without careful clinical evaluation, these individuals may be incorrectly diagnosed with PAP-A.

The prognosis for an individual with PHS and no known family history of PHS is based on the malformations present in the individual. Literature surveys are not useful for the purpose of establishing the prognosis because reported cases tend to show bias of ascertainment to more severe involvement. Although PHS has been categorized as a member of the CAVE (**c**erebro**a**cro**v**isceral **e**arly lethality) group of disorders, few affected individuals have an early lethality phenotype. This early lethality is most likely attributable to panhypopituitarism that is caused by pituitary or hypothalamic dysplasia or severe airway malformations such as laryngotracheal clefts. In addition, imperforate anus can cause serious complications if not recognized promptly. Thus, in the absence of life-threatening malformations, the prognosis should be assumed to be excellent for individuals with the nonfamilial occurrence of PHS. For individuals with a family history of affected family members, the prognosis is based on the degree of severity present in the family.

Hypothalamic hamartoma. Hypothalamic hamartoma is a malformation, not a tumor. Hypothalamic hamartomas grow at the rate of, or slower than, the surrounding brain tissue. Hypothalamic hamartomas may be large (up to 4 cm in greatest dimension); little correlation exists between the size of the hypothalamic hamartoma and presence or severity of symptoms. Individuals with hypothalamic hamartomas may have neurologic symptoms, although most are asymptomatic. Removal of the hypothalamic hamartoma is not indicated and often results in iatrogenic pituitary insufficiency.

Neurologic findings. The best-described neurologic complication of hypothalamic hamartoma is gelastic epilepsy, a partial complex seizure manifest by clonic movements of the chest and diaphragm that simulate laughing. Other types of seizures may be caused by hypothalamic hamartoma. Seizures associated with hypothalamic hamartoma in individuals with PHS are generally milder and are responsive to treatment, in contrast to individuals with nonsyndromic hypothalamic hamartoma who often have refractory seizures [Boudreau et al 2005]. No individual with PHS has been shown to have visual field loss even with a hypothalamic hamartoma near the optic chiasm.

Psychiatric and neuropsychological findings. Some individuals with PHS have behavioral manifestations including a few with severe mental retardation and behavioral disturbances [Ng et al 2004]. A larger study of behavioral manifestations of this disorder was inconclusive, reflecting the difficulty of assessing mild behavioral phenotypes in rare disorders [Azzam et al 2005].

Endocrine manifestations. The endocrine manifestations of a hypothalamic hamartoma range from isolated growth hormone deficiency or isolated precocious puberty to panhypopituitarism, which can be life threatening. Cortisol deficiency can occur in nonfamilial cases, but appears to be rare in familial cases.

Epiglottic abnormalities. Bifid epiglottis is nearly always asymptomatic; however, the more severe clefts of the larynx reported in individuals with PHS can cause severe airway symptoms. Posterior laryngeal clefts can be fatal.

Genotype-Phenotype Correlations

Genotype-phenotype correlations of *GLI3* mutations

- **GCPS** is caused by the following:
  - Actual or functional haploinsufficiency for *GLI3*
  - Truncating mutations within and 5' of the zinc finger domains and in the 3'-most third of the gene
- **PHS** is generally caused by truncating mutations in the middle third of the gene [Johnston et al 2005].

Penetrance

No instances of incomplete penetrance of PHS have been published.

Ng et al (2004) reported one individual with apparent germline mosaicism without evident clinical features.

Anticipation

Anticipation is not seen in PHS.

Nomenclature

Other descriptors used include the following:

- **Hypothalamic hamartoblastoma syndrome.** This is incorrect as "blastoma" refers to tissues in which the neural elements of hamartomas are immature, and it is also incorrect as it does not reflect the syndromic nature of the phenotype and may be confused with isolated hamartomas.
- **CAVE complex** (**c**erebro**a**cro**v**isceral **e**arly lethality). This designation is inappropriate as most individuals are mildly affected and do not manifest early lethality.
- **Hall-Pallister syndrome**

Note: The abbreviation "HPS" is used for the Hermansky-Pudlak syndrome.

Prevalence

PHS is rare. The prevalence is unknown. Approximately 100 cases are known to the author [Biesecker, personal observation]. It is suspected that many individuals with postaxial polydactyly and asymptomatic hypothalamic hamartoma or bifid epiglottis may be misdiagnosed as having nonsyndromic PAP-A.

PHS is pan ethnic.

Differential Diagnosis

For current information on availability of genetic testing for disorders included in this section, see GeneTests Laboratory Directory. —ED.

Central polydactyly

- **Oral-facial-digital syndrome type 6** includes central polydactyly with hypoplasia of the cerebellar vermis. Renal agenesis and dysplasia have been described.
- **Holzgreve syndrome** (central polydactyly, cleft palate, heart defect)

Postaxial polydactyly

- **McKusick-Kaufman syndrome (MKS)** is characterized by the triad of hydrometrocolpos in females and genital malformations in males, postaxial polydactyly (PAP) or central polydactyly, and congenital heart disease (CHD). Mutations in the *MKKS* gene have been found in individuals with MKS within the Amish population. Inheritance is autosomal recessive.
- **Holt-Oram syndrome (HOS)** is characterized by upper-extremity malformations involving radial, thenar, or carpal bones and a personal and/or family history of congenital heart malformation, most commonly ostium secundum atrial septal defect (ASD) and ventricular septal defect (VSD), especially those occurring in the muscular trabeculated septum and/or cardiac conduction disease. *TBX5* is the only gene currently known to be associated with HOS. *TBX5* mutations are found in more than 70% of individuals with HOS. Inheritance is autosomal dominant.
- **Bardet-Beidl syndrome** is characterized by rod-cone dystrophy, truncal obesity, postaxial or central polydactyly, cognitive impairment, male hypogonadotrophic hypogonadism, complex female genitourinary malformations, and renal dysfunction, which is a major cause of morbidity and mortality. Twelve genes are known to be associated with Bardet-Biedl syndrome: *BBS1*, *BBS2*, *ARL6*, *BBS4*, *BBS5*, *MKKS*, *BBS7*, *TTTC8*, *BBS9*, *BBS10*, *TRIM32*, and *BBS12*. Inheritance is autosomal recessive. Hypothalamic hamartoma and bifid epiglottis are rare manifestations of Bardet-Biedl syndrome [Stevens & Ledbetter 2005].
- **Smith-Lemli-Opitz syndrome (SLOS)** is a congenital multiple anomaly syndrome caused by an abnormality in cholesterol metabolism resulting from deficiency of the enzyme 7-dehydrocholesterol reductase. It is characterized by prenatal and postnatal growth retardation, microcephaly, moderate-to-severe mental retardation, and multiple major and minor malformations including postaxial polydactyly. *DHCR7* is the only gene known to be associated with SLOS. Inheritance is autosomal recessive.

Hypothalamic hamartoma. Nonsyndromic or isolated hypothalamic hamartomas may cause either endocrine disturbance (most commonly, growth hormone deficiency or precocious puberty) or a severe neurologic picture of refractory seizures, behavior problems, and cognitive decline. Gelastic epilepsy may be associated.

Management

Evaluations Following Initial Diagnosis

To establish the extent of disease in an individual diagnosed with Pallister-Hall syndrome (PHS), the following evaluations are recommended:

- Assessment for cortisol deficiency. This must be performed urgently in individuals who have no family history of PHS and in individuals who have family members with PHS and cortisol deficiency. Of note, adrenal crisis can be lethal in infants who have not undergone proper evaluation and treatment for adrenal insufficiency.
- Consultation by an endocrinologist, including evaluation of growth hormone secretion, FSH and LH secretion, and serum concentration of thyroid hormone in early infancy after evaluation for and treatment of ACTH deficiency

- Cranial MRI to establish the location and extent of hamartoma
- Neurologic examination to exclude signs of intracranial hypertension, which is not typical of hypothalamic hamartomas
- Limb radiographs to distinguish postaxial polydactyly from central polydactyly
- Renal ultrasonography to look for renal anomalies
- Visualization of the epiglottis by laryngoscopy. Urgent evaluation by an otolaryngologist for laryngotracheal cleft when signs or symptoms of aspiration are present. Elective evaluation by an otolaryngologist in asymptomatic individuals for the purpose of establishing the diagnosis or establishing the extent of anomalies.
- Surgical consultation for imperforate anus or anal stenosis if present
- Developmental assessment

Treatment of Manifestations

Endocrine abnormalities are treated as in the general population, with treatment for cortisol deficiency being the most urgent.

Anal atresia or stenosis should be treated in standard fashion.

Management of epiglottic abnormalities depends on the type of abnormality and extent of respiratory compromise and is the same as in the general population. Bifid epiglottis is commonly asymptomatic and most do not require treatment, unless accompanied by clear evidence of obstruction or associated with other anomalies, such as tracheal stenosis.

Seizures are treated symptomatically. Seizures associated with PHS are commonly responsive to antiepileptic drugs (AEDs), whereas seizures associated with nonsyndromic hypothalamic hamartomas are more commonly refractory to AEDs.

Repair of polydactyly should be undertaken on an elective basis.

If developmental delays are detected, intervention and/or special education are indicated.

Prevention of Secondary Complications

Only under the most unusual circumstances should a hypothalamic hamartoma be removed or even biopsied because the complications of surgery and the need for lifelong hormone supplements postoperatively generally outweigh the benefits.

Use of stimulants for attention deficit disorder should be considered carefully in persons with a CNS lesion that predisposes to seizures (e.g., hypothalamic hamartoma).

Surveillance

During childhood:

- Annual medical evaluations to assess growth and monitor for signs of precocious puberty
- Annual screening for developmental delay or learning disorders

Testing of Relatives at Risk

See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes.

Therapies Under Investigation

Search ClinicalTrials.gov for access to information on clinical studies for a wide range of diseases and conditions. Note: There may not be clinical trials for this disorder.

Other

Genetics clinics are a source of information for individuals and families regarding the natural history, treatment, mode of inheritance, and genetic risks to other family members as well as information about available consumer-oriented resources. See the GeneTests Clinic Directory.

Support groups have been established for individuals and families to provide information, support, and contact with other affected individuals. The Resources section (below) may include disease-specific and/or umbrella support organizations.

Genetic Counseling

Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members. This section is not meant to address all personal, cultural, or ethical issues that individuals may face or to substitute for consultation with a genetics professional. To find a genetics or prenatal diagnosis clinic, see the GeneTests Clinic Directory.

Mode of Inheritance

Pallister-Hall syndrome (PHS) is inherited in an autosomal dominant manner.

Risk to Family Members

Parents of a proband

- Some individuals diagnosed with PHS have an affected parent and some have the disorder as the result of a *de novo* gene mutation. The proportion of cases caused by *de novo* gene mutations is unknown, as the frequency of subtle signs of the disorder in parents has not been thoroughly evaluated and molecular genetic data are insufficient.
- Because all affected parents to date have had polydactyly, the parents of a proband with no known family history of PHS should be examined for evidence of extra digits.

Sibs of a proband

- The risk to the sibs of a proband depends on the status of the parents of the proband.
- If a parent is affected, the risk to the sibs is 50%.
- When the parents are clinically unaffected, the risk to the sibs of a proband appears to be low, but greater than that of the general population because of the possibility of germline mosaicism. One instance of parental mosaicism has been reported [Ng et al 2004].

  Note: The description of a single case of mosaicism for a rare disorder does not allow an estimation of the frequency of this event for genetic recurrence risk estimates, but it must be considered as a possibility.

Offspring of a proband

- Every child of an individual with PHS has a 50% chance of inheriting the mutation.

- Becausef intrafamilial variability appears to be low, affected offspring would be expected to have clinical findings similar to those of the parent.

Other family members of a proband. The risk to other family members depends on the status of the proband's parents. If a parent is found to be affected or to have a disease-causing mutation, his or her family members are at risk.

Related Genetic Counseling Issues

Considerations in families with an apparent *de novo* mutation. When neither parent of a proband with an autosomal dominant condition has the disease-causing mutation or clinical evidence of the disorder, it is likely that the proband has a *de novo* mutation. However, possible non-medical explanations including alternate paternity or maternity (i.e., with assisted reproduction) or undisclosed adoption could also be explored.

Family planning. The optimal time for determination of genetic risk is before pregnancy. It is appropriate to offer genetic counseling (including discussion of potential risks to offspring and reproductive options) to young adults who are affected.

DNA banking. DNA banking is the storage of DNA (typically extracted from white blood cells) for possible future use. Because it is likely that testing methodology and our understanding of genes, mutations, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals. DNA banking is particularly relevant when the sensitivity of currently available testing is less than 100%. See DNA Banking for a list of laboratories offering this service.

Prenatal Testing

Molecular genetic testing. Prenatal diagnosis for pregnancies at increased risk is possible by analysis of DNA extracted from fetal cells obtained by amniocentesis usually performed at approximately 15-18 weeks' gestation or chorionic villus sampling (CVS) at approximately ten to 12 weeks' gestation. The disease-causing allele of an affected family member must be identified before prenatal testing can be performed.

Note: Gestational age is expressed as menstrual weeks calculated either from the first day of the last normal menstrual period or by ultrasound measurements.

Ultrasound examination. In fetuses at 50% risk, prenatal ultrasound examination may detect polydactyly. However, a normal ultrasound examination does not eliminate the possibility of PHS in the fetus.

Preimplantation genetic diagnosis (PGD) may be available for families in which the disease-causing mutation has been identified. For laboratories offering PGD, see Testing.

Molecular Genetics

Information in the Molecular Genetics tables is current as of initial posting or most recent update. —ED.

Table A. Molecular Genetics of Pallister-Hall Syndrome

Gene Symbol	Chromosomal Locus	Protein Name
GLI3	7p13	Zinc finger protein GLI3

Data are compiled from the following standard references: Gene symbol from HUGO; chromosomal locus, locus name, critical region, complementation group from OMIM; protein name from Swiss-Prot.

Table B. OMIM Entries for Pallister-Hall Syndrome

146510	PALLISTER-HALL SYNDROME; PHS
165240	GLI-KRUPPEL FAMILY MEMBER 3; GLI3

Table C. Genomic Databases for Pallister-Hall Syndrome

Gene Symbol	Entrez Gene	HGMD
GLI3	2737 (MIM No. 165240)	GLI3

For a description of the genomic databases listed, click here.

Note: HGMD requires registration.

Normal allelic variants: The *GLI3* gene extends over approximately 276 kb and includes 15 exons. The mRNA is approximately 8 kb, the reference cDNA is 8,209 bp (NM_000168.3, NP_000159.3), and the open reading frame is 4,740 bp.

A number of putative normal allelic variants exist in *GLI3* (Table 2; pdf). Most of the variants have been seen in multiple unrelated persons and are not believed to be associated with any phenotypic effects, although they have not been rigorously analyzed for subtle effects. They are included in Table 2 if they lie within an exon or if they are in an intron within 25 bp of an exon. Readers should refer to dbSNP to confirm these data and for additional data. (SNPs are from Human Genome build 126).

Pathologic allelic variants: Selected pathologic variants reported in individuals with PHS are in Table 3 (pdf). Multiple new mutations have been identified by Johnston et al (2005).

Normal gene product: The gene encodes a protein of 1,580 amino acids.

Note: As the result of a cDNA sequencing error, older citations described a longer open reading frame that predicted a protein of 1,596 amino acids; the error has been corrected in the GenBank entry NM_000168.3.

Abnormal gene product: It is hypothesized that truncated forms of the GLI3 protein repress transcription.

Resources

GeneReviews provides information about selected national organizations and resources for the benefit of the reader. GeneReviews is not responsible for information provided by other organizations. Information that appears in the Resources section of a GeneReview is current as of initial posting or most recent update of the GeneReview. Search GeneTests*for this disorder and select* Resources *for the most up-to-date Resources information.*—ED.

National Library of Medicine Genetics Home Reference
Pallister-Hall syndrome

American Epilepsy Society
342 North Main Street
West Hartford CT 06117-2507
Phone: 860-586-7505
Fax: 860-586-7550
Email: info@aesnet.org
www.aesnet.org

Epilepsy Foundation
8301 Professional Place
East Landover, MD 20785-2238
Phone: 800-EFA-1000 (800-332-1000); 301-459-3700
Fax: 301-577-4941
Email: webmaster@efa.org
www.efa.org

Medline Plus
Polydactyly

References

Medical Genetic Searches: A specialized PubMed search designed for clinicians that is located on the PubMed Clinical Queries page. PubMed

Published Statements and Policies Regarding Genetic Testing

No specific guidelines regarding genetic testing for this disorder have been developed.

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Suggested Readings

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Chapter Notes

Author Notes

Author's Web page

The author is a board-certified clinical geneticist and pediatrician. He performs clinical and molecular research on PHS and related disorders at the NIH.

Revision History

- 18 March 2008 (me) Comprehensive update posted to live Web site
- 2 June 2006 (cd) Revision: prenatal diagnosis clinically available
- 6 June 2005 (me) Comprehensive update posted to live Web site
- 1 May 2003 (me) Comprehensive update posted to live Web site
- 25 May 2000 (me) Review posted to live Web site
- 20 January 2000 (lb) Original submission