Townes-Brocks Syndrome

Townes-Brocks syndrome is a genetic condition that affects several parts of the body. The most common features of this condition are an obstruction of the anal opening (imperforate anus), abnormally shaped ears, and hand malformations that most often affect the thumb. Most people with this condition have at least two of these three major features.

Other possible signs and symptoms of Townes-Brocks syndrome include kidney abnormalities, mild to profound hearing loss, heart defects, and genital malformations. These features vary among affected individuals, even within the same family. Intellectual disability or learning problems have also been reported in about 10 percent of people with Townes-Brocks syndrome.

Frequency

The prevalence of this condition is unknown, although one study estimated that it may affect 1 in 250,000 people. It is difficult to determine how frequently Townes-Brocks syndrome occurs because the varied signs and symptoms of this disorder overlap with those of other genetic syndromes.

Causes

Mutations in the SALL1 gene cause Townes-Brocks Syndrome. The SALL1 gene is part of a group of genes called the SALL family. These genes provide instructions for making proteins that are involved in the formation of tissues and organs before birth. SALL proteins act as transcription factors, which means they attach (bind) to specific regions of DNA and help control the activity of particular genes. Some mutations in the SALL1 gene lead to the production of an abnormally short version of the SALL1 protein that malfunctions within the cell. Other mutations prevent one copy of the gene in each cell from making any protein. It is unclear how these genetic changes disrupt normal development and cause the birth defects associated with Townes-Brocks syndrome.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- anal-ear-renal-radial malformation syndrome
- deafness-imperforate anus-hypoplastic thumbs syndrome
- imperforate anus-hand and foot anomalies syndrome
• renal-ear-anal-radial syndrome (REAR)
• sensorineural deafness-imperforate anus-hypoplastic thumbs syndrome
• Townes syndrome

Diagnosis & Management

Genetic Testing Information

• What is genetic testing? /primer/testing/genetictesting

Other Diagnosis and Management Resources


Additional Information & Resources

Health Information from MedlinePlus

• Encyclopedia: Ear Disorders (image) https://medlineplus.gov/ency/imagepages/17281.htm
• Encyclopedia: Imperforate Anus https://medlineplus.gov/ency/article/001147.htm
• Health Topic: Hand Injuries and Disorders https://medlineplus.gov/handinjuriesanddisorders.html
• Health Topic: Hearing Disorders and Deafness https://medlineplus.gov/hearingdisordersanddeafness.html
• Health Topic: Kidney Diseases https://medlineplus.gov/kidneydiseases.html

Genetic and Rare Diseases Information Center

• Townes-Brocks syndrome https://rarediseases.info.nih.gov/diseases/7784/townes-brocks-syndrome
Educational Resources

- American Society for Surgery of the Hand: Congenital Hand Differences
  http://handcare.assh.org/Anatomy/Details-Page/articleId/39392
- Cincinnati Children's Hospital Medical Center: Anorectal Malformations
  https://www.cincinnatichildrens.org/health/a/anorectal-malformations
- MalaCards: townes-brocks syndrome
  https://www.malacards.org/card/townes_brocks_syndrome
- Orphanet: Townes-Brocks syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=857

Patient Support and Advocacy Resources

- National Association of the Deaf
  https://www.nad.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/townes-brocks-syndrome/
- Resource list from the University of Kansas Medical Center: Deafness and Hard of Hearing
  http://www.kumc.edu/gec/support/hearing.html

Clinical Information from GeneReviews

- Townes-Brocks Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1445

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28townes-brocks+syndrome%5BTIAB%5D%29+OR+%28townes+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- TOWNES-BROCKS SYNDROME 1
  http://omim.org/entry/107480
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17221874

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16089222

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11478532

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12915476

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301618

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10051003
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1734298/

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