

DISEASE:
Townes-Brocks syndrome

NAME:	Townes-Brocks syndrome
DESCRIPTION:	A rare genetic disorder characterized by the triad of imperforate anus, dysplastic ears often associated with sensorineural and/or conductive hearing impairment, and thumb malformations. These features are often associated with other signs mainly affecting the kidneys and heart.
ORPHACODE:	857
SYNONYMS:	Imperforate anus-hand, foot and ear anomalies syndrome REAR syndrome Renal-ear-anal-radial syndrome Sensorineural deafness with imperforate anus and hypoplastic thumbs Sensorineural hearing loss with imperforate anus and hypoplastic thumbs TBS Townes syndrome
XREF(S):	Orphanet OMIM MeSH OMIM ICD-10

ANALYTE(S):	<u>SALL1</u> <u>DACT1</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/164>

RELATED CONTENT

Related Genetic Tests

- [Renal or urinary tract malformation \(CAKUT\) \(gene panel\)](#)

Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)

Related Analytes

- [dishevelled binding antagonist of beta catenin 1](#)
- [spalt like transcription factor 1](#)

Related Gene Panels

- [Cakut \(congenital anomalies of the kidney and urinary tract-1\) \(69 genes\) - IPG](#)

Source URL: <http://gentest.healthdata.be/disease/164>