
Lab Dept: Anatomic Pathology

Test Name: TOWNES-BROCKS SYNDROME (SALL1)
SEQUENCING

General Information

Lab Order Codes: SALL

Synonyms: TBS; Renal-Ear-Anal-Radial (REAR) syndrome; Imperforate anus with hand, foot, and ear anomalies; Sensorineural deafness with imperforate anus and thumb anomalies

CPT Codes: 81479 – Molecular Pathology Unlisted procedure

Test Includes: Analysis is performed bi-directional sequencing of the coding regions (exons 1-3) and splice sites of the SALL1 gene. Mutations found in the first of a family to be tested and is confirmed by repeat analysis using sequencing, restriction fragment analysis, or another appropriate method.

Logistics

Test Indications: Townes Brocks syndrome is a rare multiple malformation syndrome characterized by anal, limb, ear, and renal anomalies. Intelligence is normal in most affected individuals. Diagnostic features include ano-rectal abnormalities (imperforate or anteriorly placed anus, anal stenosis, prominent midline perineal raphe); abnormalities of the hands and feet (preaxial polydactyly, triphalangeal thumbs, bifid thumbs and toes, finger and toe syndactyly); external ear malformations (preauricular tags or pits, "loop" or "satyr" ear, microtia, abnormal helix) with hearing loss (sensorineural, conductive or mixed); and renal abnormalities leading to impaired renal function or renal failure (unilateral or bilateral hypoplastic or dysplastic kidneys, multicystic kidneys, renal agenesis, posterior urethral valves, vesico-uretal reflex). Other, less common features are cardiac defects, mental retardation, eye, genitourinary and vertebral abnormalities, hypothyroidism, umbilical hernia, and gastroesophageal reflux. The intra- and interfamilial clinical presentation of TBS varies widely and overlaps with several other disorders including VATER and VACTERL associations, Okihiro syndrome, Fanconi anemia, Baller-Gerold syndrome, branchio-oto-renal (BOR) syndrome and oculo-auriculo-vertebral (OAV) spectrum. Important differentiating characteristics of TBS are the absence of radial hypoplasia, craniostenosis, and tracheo-esophageal fistula.

Reasons for referral:

1. Confirmation of a clinical diagnosis
2. Genetic counseling and risk assessment

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDx Test: 2521)

| | |
|------------------------------|---|
| Phone Numbers: | MIN Lab: 612-813-6280 STP Lab: 651-220-6550 |
| Test Availability: | Daily, 24 hours. Specimens collected Saturday or Saturday will be held for shipment on Monday. |
| Turnaround Time: | 3 weeks |
| Special Instructions: | A GeneDx signed request form must be sent with any patient or specimen to the laboratory. |

Specimen

| | |
|-----------------------------|---|
| Specimen Type: | Whole blood |
| Container: | Lavender top (EDTA) tube |
| Draw Volume: | 2 - 5 mL (Minimum: 1 mL) blood |
| Processed Volume: | Same as Draw Volume |
| Collection: | Routine venipuncture for blood specimens, invert gently to mix |
| Special Processing: | Lab Staff: Send whole blood in original collection container labeled with patient name, date of birth and medical record number, including signed consent form and requisition, with a cool pack in warm weather, via overnight or second-day courier so that the sample will arrive at GeneDx, Monday through Saturday. Samples drawn on Saturday or Sunday should be held at refrigerated temperatures for shipment on Monday. Do not freeze. Note: Specimens may be stored at refrigerated temperatures for up to 7 days prior to shipping. |
| Patient Preparation: | None |
| Sample Rejection: | Unrefrigerated specimens older than 48 hours; clotted or hemolyzed for blood; frozen specimens; mislabeled or unlabeled specimens |

Interpretive

| | |
|-------------------------|-----------------------|
| Reference Range: | No mutations detected |
| Critical Values: | N/A |
| Limitations: | N/A |
| Methodology: | Capillary sequencing |

References:

[GeneDx, Inc.](#) February 2018
(301)519-2100 Fax (301)519-2892

Updates:

2/11/2013: CPT update