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 charateristics 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#: 2521)

Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours (Preferred draws are Sunday - Thursday as specimens can only be received at the reference lab Monday - Friday. Specimens collected Friday or Saturday will be held for shipment on Monday.)
Turnaround Time:	8 weeks for new patients
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:	1 - 5 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, Inc. on a weekday (Monday through Friday). Samples drawn on Friday or Saturday should be held at refrigerated temperatures for shipment on Sunday. **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:	Buccal swabs are not an acceptable specimen for this testing.
Methodology:	Bi-directional sequence analysis

References: GeneDx, Inc. March 2012
(301)519-2100 Fax (301)519-2892

Updates: 2/11/2013: CPT update