
Lab Dept: Anatomic Pathology

Test Name: SHWACHMAN DIAMOND SYNDROME (SBDS) SEQUENCING

General Information

Lab Order Codes: SBD (Blood or Buccal Swab)

Synonyms: Shwachman-Bodian Syndrome; Shwachman-Bodian-Diamond Syndrome; SDS; Pancreatic insufficiency and bone marrow dysfunction; congenital lipomatosis of the pancreas

CPT Codes: 81479 – Molecular Pathology Unlisted procedure

Test Includes: Analysis is performed by bi-directional sequencing of the coding regions and splice sites of exons 1-5 of the SBDS 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: Shwachman-Diamond syndrome (SDS) is an autosomal recessive disorder that includes pancreatic exocrine insufficiency and hematological abnormalities as consistent features. Other common manifestations include skeletal abnormalities, short stature, liver dysfunction and increased risk of malignancy. Serious infections and acute myeloid leukemia are major causes of mortality and morbidity. The syndrome is caused by the partial, not complete, deficiency of the novel protein encoded by the SBDS gene, thought to be involved in RNA metabolism.

In most studies 75-89% of patients with Shwachman-Diamond Syndrome have at least one SBDS gene mutation detected, and usually two. No other gene is known to cause this syndrome. Detection of one mutation is suggestive of the diagnosis, but not definitive. Detection of two mutations is usually definitive but because in this gene two mutations sometimes occur side by side in the same copy (allele) of the gene, follow-up parental testing may be necessary.

Reasons for referral:

1. Confirmation of clinical diagnosis.
2. Confirmation that a potential hematopoietic stem cell transplant donor is not affected.
3. Genetic counseling/ or carrier testing in relatives.
4. Pre-natal diagnosis in at-risk pregnancies.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx, Inc. (GDX#: 109)

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: 6 weeks for new patients

Special Instructions: A GeneDx signed [request form](#) must be sent with any patient or specimen to the laboratory.

SBDS can be tested using cells obtained by swabbing the buccal mucosa (inside of cheek). Buccal specimens are not accepted on children under 6 months of age. Buccal swab collection kits are available from GeneDx (ph. 301-519-2100).

Specimen

Specimen Type: Whole blood
Buccal cell swab (preferred specimen)

Container: Blood: Lavender top (EDTA) tube
Buccal Smear: Cytobrush Plus Cell Collector® kit

Draw Volume: 1 - 5 mL blood
2 swabs from the Cytobrush Plus Cell Collector® kit

Processed Volume: Same as Draw Volume

Collection: Routine venipuncture for blood specimens, invert gently to mix
Buccal swab kit for buccal swabs:

Buccal Cell Collection procedure:

1. Remove a swab from the Buccal swab kit touching only the “stick” end.
2. **Do Not** rinse mouth before starting. Have the individual open his/her mouth. Twirl the swab on the inner cheek for 30 seconds. **Do Not** scrape so hard that the cheek bleeds.
3. Place the swab end in the labeled tube. Cut the “stick” with scissors at the level of the top of the tube. Replace cap and close completely.
4. Repeat the process with another swab on the opposite cheek.
5. Send the kit to the lab for processing and mailing.

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.

Mail the Cytobrush Plus Cell Collector® kit, including signed consent form and requisition, by regular mail to GeneDx in the included envelope.

Patient Preparation: For buccal cell collection, **Do Not** have the patient rinse his/her mouth.

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 specimens are not accepted on children under 6 months of age.

Methodology: Bi-directional sequence analysis

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

Updates: 2/11/2013: CPT update