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**Lab Dept:**                   **Anatomic Pathology**

**Test Name:**               **SHWACHMAN DIAMOND SYNDROME (SBDS)  
TWO KNOWN MUTATIONS**

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***General Information***

**Lab Order Codes:**       SB2K (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:**           Testing of a relative for two specific known mutations (carrier testing) where testing was previously done by GeneDx. Using genomic DNA, the exon or exons of interest are screened by bi-directional sequence analysis and/or by non-sequence methods such as heteroduplex analysis or restriction enzyme digestion. The previously tested proband DNA serves as a positive control.

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***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.

**Carrier testing reasons for referral:**

1. Testing parents of a child with a specific dominant mutation, in order to determine if the mutation in the child is new or inherited.
2. Carrier testing in parents of a child with apparently homozygous recessive mutations, to rule out the possibility that the child has one mutated allele and one allele that is deleted or refractory to amplification.
3. Carrier testing in the parents of a child with recessive mutations, to confirm that all four parental alleles can be detected prior to prenatal diagnosis.
4. Pre-symptomatic testing in siblings of the index case.
5. Carrier testing in the extended family.

**Lab Testing Sections:**   Anatomic Pathology - Sendouts

**Referred to:**             GeneDx, Inc. (GDx#: 9012, Specify gene/two mutations)

**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:** 3-4 weeks

**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).

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### ***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

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### ***Interpretive***

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** Buccal specimens are not accepted on children under 6 months of age.

**Methodology:** Bi-directional sequence analysis or other methods as required

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

**Updates:** 2/11/2013: CPT update