RESEARCH OPPORTUNITY FOR INDIVIDUALS WITH SUSPECTED SHWACHMAN-DIAMOND SYNDROME

ABOUT THE PROJECT

The Rare Genomes Project is a free and remote research program using genomic sequencing to look for the genetic cause of rare diseases, such as Shwachman-Diamond Syndrome. Eligible families will be asked to provide a blood sample and medical information. If a result is found, we will work with your doctor to confirm the result.

This research process takes longer than routine genetic testing and not all families will have a result identified.

ELIGIBLE PARTICIPANTS

- Have a clinical suspicion for Shwachman-Diamond Syndrome, including a history of **two or more** of the following symptoms:

  Exocrine pancreatic insufficiency
  Decreased pancreatic enzymes (serum trypsinogen or pancreatic isoamylase), decreased fecal elastase, malabsorption, or steatorrhea

  Hematologic abnormalities
  Cytopenias, hypocellular bone marrow, bone marrow failure, or MDS/AML

  Skeletal dysplasia
  Rib cage/thoracic abnormality, metaphyseal dysostosis, extremity abnormalities, scoliosis, or abnormal bone density

- Have a suspected genetic cause that has **not** been identified due to prior testing being negative or inconclusive **OR** a lack of access to genetic testing
- Live in the United States

LEARN MORE AND APPLY

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SHWACHMAN-DIAMOND SYNDROME ALLIANCE

rare genomes project