Pearson Marrow-Pancreas Syndrome: Mitochondrial Deletions/Duplications

Test Code: ZN
Turnaround time: 4 weeks
CPT Codes: 81401 x1

Condition Description

Mitochondrial DNA (mtDNA) deletions result in a spectrum of multi-system disorders that include: Kearns-Sayre Syndrome (KSS), Pearson Syndrome, and Progressive External Ophthalmoplegia (CPEO).

Pearson Syndrome, usually fatal in infancy, is a severe manifestation of mtDNA deletion that is characterized by anemia and pancreatic dysfunction. Pearson Syndrome patients present early in life with a severe transfusion-dependent, macrocytic anemia with varying degrees of neutropenia and thrombocytopenia. The bone marrow shows extensive vacuolization of erythroid and myeloid precursors, hemosiderosis, and ringed sideroblasts. Many patients develop exocrine pancreatic insufficiency, hepatic failure, renal failure, and other neuromuscular problems. Rare Pearson syndrome patients spontaneously recover the ability to make blood cells and progress to Kearns-Sayre-like phenotype.

Pearson Syndrome caused by deletions in mtDNA usually occurs spontaneously, but in rare cases has been maternally inherited. The great majority of these rearrangements are new mutations, therefore, patients are heteroplasmic for the presence of normal and rearranged mtDNA molecules. Since mutant mtDNA molecules may be undetectable in blood cells due to replicative segregation (uneven tissue distribution of mitochondrial molecules), analysis of bone marrow may be necessary if the blood sample is negative for mtDNA rearrangements or mutations.

Please [click here](#) for the GeneReviews summary on this condition.

Indications

This test is indicated for:

- Patients with a clinical diagnosis or suspicion of Pearson Syndrome.

Methodology

Deletions in mtDNA are detectable in blood or bone marrow by Southern blot, using digestion with EcoRV, Bam H1, and SnaB1, followed by hybridization with mtDNA as a probe.

Detection

Deletions are by nature expected to be heteroplasmic and levels can vary between tissues. MitDNA deletions are almost always detectable in the bone marrow of symptomatic patients. In over 95% of cases, Pearson Syndrome has caused deletions that are detectable by Southern blot.

Reference Range

Qualitative assay.

Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Bone Marrow**

Specimen Requirements:

In EDTA (purple top) tube: 2-3 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Related Tests

- Kearns-Sayre Syndrome and Chronic Progressive External Ophthalmoplegia Assay (QB)