Peroxisome Disorders: Deletion/Duplication Panel

Test Code: MD140
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Peroxisomes are intracellular organelles with diverse cellular functions including biosynthesis, catabolism and detoxification of various compounds. Inborn errors of metabolism of peroxisomes function are roughly divided into peroxisome biogenesis disorders and single enzyme or transporter defects. This panel is designed for comprehensive testing for the following peroxisomal disorders:

1. Peroxisome biogenesis disorders, also called Zellweger syndrome spectrum (PBD, ZSS) that include Zellweger syndrome (ZS), neonatal adrenoleukodystrophy (NALD), and infantile Refsum disease (IRD).
2. X-linked adrenoleukodystrophy and adrenomyeloneuropathy.
3. Rhizomelic Chondrodysplasia Punctata, Types 1, 2, and 3.
4. Refsum Disease or phytanoyl-CoA hydroxylase deficiency.
5. Acyl-CoA oxidase (ACOX1) deficiency.
6. D-bifunctional protein (DBP) deficiency
7. 2-Methylacyl-CoA racemase (AMACR) deficiency.
8. Acatasemia.
9. Hyperoxaluria Type 1 or alanine glyoxylate aminotransferase deficiency.
10. Mullibrey nanism.
11. Sterol carrier protein X (SCP2) deficiency.
12. DNM1L-related encephalopathy.
13. ABCD3-related congenital bile acid synthesis defect

Reference:

- GeneReviews.

Genes

ABCD1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7

Indications

This test is indicated for:

- Confirmation of a clinical/biochemical diagnosis of peroxisomal disorders.
- Carrier testing in adults with a family history of peroxisomal disorders.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient’s phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient’s clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood

Specimen Requirements:

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

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

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

### Related Tests

- Peroxisome Disorders: Sequencing Panel
- Individual gene sequencing and deletion/duplication analysis are available for the following genes: ABCD1, PEX1, PEX2, PEX3, PEX5, PEX6, PEX12, PEX14, and PEX26