Endocrine Disorders: Deletion/Duplication Panel

Test Code: MD290
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

The Endocrine Disorders Panel primarily tests for two broad categories of endocrine disorders: (1) disorders of sexual development (DSD) and hormone production, and (2) transient or permanent neonatal diabetes mellitus (NDM) and maturity onset diabetes of the young (MODY). DSD may manifest in infancy with ambiguous genitalia or at puberty when atypical sexual development occurs. DSD include such things as hypogonadotropic hypogonadism (with or without anosmia), premature ovarian failure or ovarian dysgenesis, and congenital adrenal hyperplasia. DSD are important to diagnose early for proper treatment and management of these conditions. NDM and MODY are genetically heterogeneous disorders. A molecular genetic diagnosis is critical, since some monogenic diabetes can be treated with sulfonylureas, instead of requiring life-long insulin therapy.

References:

- Hughes et al., 2013 *Endocrine* abstracts 33: P48.

Genes

- ABCB6, AGPAT2, AKT2, BLK, BMP15, BSCL2, CHD7, CIDE3, CISD2, CYP17A1, CYP19A1, EIF2AK3, FGFR8, FGFR1, FIGLA, FOXP3, FSHR, GATA4, GCK, GDF9, GLIS3, GNRH1, GNRSF, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KISS1, KISS1R, KLF11, LHCG, LMNA, NEUROD1, NOBOX, NRB1, NR5A1, NIS, PAX4, PDX1, POU1F1, PPARG, PROK2, PROKR2, PSMB3IP, PTF1A, PTRF, RFX6, SLC2A2, TAC3, TACR3, TBC1D4, WFS1, ZMPSTE24

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of disorders of sexual development.
- Confirmation of a clinical diagnosis of disorders of hormonal imbalance.
- Confirmation of a clinical diagnosis of neonatal diabetes mellitus.
- Confirmation of a clinical diagnosis of mature onset diabetes of the young.

Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which 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

- Endocrine Disorders: Sequencing Panel
- Maturity Onset Diabetes of the Young Panel