Lab Dept: Anatomic Pathology

Test Name: KALLMAN SYNDROME (KAL1) GENE SEQUENCING

General Information

Lab Order Codes: KAL1G

Synonyms: EDS IV, COL3A1; Callman syndrome (KS) sequencing; Hypogonatropic hypogonadism; Anosmia

CPT Codes: 81406 – Molecular Pathology, Level 7

Test Includes: Bi-directional sequence of the coding region and splice junctions.

Logistics

Test Indications: Kallmann syndrome (KS) is typically characterized by hypogonadotrophic hypogonadism and anosmia. The presence of a defective sense of smell, whether partial (hyposmia) or complete (anosmia) distinguishes KS from normosmic idiopathic hypogonadotropic hypogonadism with a normal sense of smell (nIHH), which can be associated with mutations in the GnRHR and GPR54 genes. Due to hypothalamic GnRH deficiency, males with KS demonstrate cryptorchidism, testicular atrophy and microphallus at birth and then subsequent failure to undergo a normal puberty during adolescence. Females with KS usually present with primary amenorrhea or infertility. Mutations in at least two genes have been shown to be associated with KS. Mutation in the X-linked KAL1 gene is associated with the classic genital and olfactory features of Kallmann syndrome, and in some cases, renal agenesis in males only. This is known as Type 1 Kallmann syndrome (KAL1). Type 1 KS is the result of abnormal anosmin-1 protein production due to KAL1 gene mutation.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: GeneDx (GeneDx Test: 2401)

Phone Numbers: MIN Lab: 612-813-6280
STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 8 – 10 weeks
**Special Instructions:**
A completed GeneDx request form is required (select test 2401 under Other Genetic Disorders). Please submit requisition with specimen or patient sent to the laboratory.

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### Specimen

**Specimen Type:** Whole blood

**Container:** Lavender top (EDTA) tube

**Draw Volume:** 5 mL (Minimum: 1 mL) blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine venipuncture

**Special Processing:** Lab Staff: *Do Not* centrifuge. Specimen should be sent in original collection container. Store and ship at refrigerated temperatures. Forward promptly.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimen; inappropriate specimen type

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### Interpretive

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** N/A

**Methodology:** Bi-directional Sequence Analysis

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

**Updates:**
- 2/6/2013: CPT update
- 3/26/2014: CPT update