
Lab Dept: Anatomic Pathology

Test Name: KALLMAN SYNDROME (KAL1) GENE SEQUENCING

General Information

Lab Order Codes: KAL1G

Synonyms: EDS IV, COL3A1; Kallman syndrome (KS) sequencing; Hypogonotropic 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 hypogonadotropic 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.

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

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