Lab Dept:	**Anatomic Pathology**
Test Name:	**X-LINKED HYPOPHOSPHATEMIA (PHEX) SEQUENCING**

General Information

Lab Order Codes:	PHEX
Synonyms:	Hypophosphatemic rickets; Vitamin D-resistant rickets
CPT Codes:	81406 – Molecular Pathology, Level 7
Test Includes:	22 coding exons and splice junctions of the PHEX gene are screened by bi-directional sequence analysis.

Logistics

Test Indications:	Use to confirm the clinical diagnosis; differentiate between X-linked and dominant forms of the disease; determine the appropriate therapeutic approach. X-linked hypophosphatemia is the most common form of inherited rickets. Affected individuals have hypophosphatemia due to decrease renal tubular serum inorganic phosphorous reabsorbtion. It is resistant to treatment with Vitamin D. The disorder includes short stature, bowing of the lower limbs, poor dental development and extra-skeletal ossification. Occasionally, spinal cord compression is present. As an X-linked dominant trait, this disorder affects both males and female. The disease is equally severe in young boys and girls, although adult males may be more several affected than adult females. A less common form of dominant hypophosphatemia that is phenotypically similar to the X-linked form is due to gain-of-function mutation in the autosomal FGF23 gene.
Lab Testing Sections:	Anatomic Pathology – Sendouts
Referred to:	GeneDx (GDX Test: 1861)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	Results are reported within 3 weeks

Special Instructions:	A complete GeneDx request form is required (select test 1861 under Hereditary rickets section). Please submit requisition with the specimen to the laboratory.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	2-5 mL (Minimum: 1mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not Centrifuge. Blood specimen should remain in the original collection container. Store and ship blood specimens at **refrigerated temperature.** Specimen may be stored refrigerated up to 7 days before shipping.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	Multiple partial gene deletions have been reported in PHEX, which would not be detectable in carrier females by gene sequencing. Therefore, GeneDX performs deletion/duplication analysis in females concurrently with sequencing to evaluate for a whole or partial gene deletion in PHEX.
Methodology:	Capillary sequencing
References:	GeneDx February 2018 301-519-2100 Fax: 301-519-2892
Updates:	2/11/2013: CPT update 3/26/2014: CPT update