Fanconi Anemia, group A, FANCA, Deletion/Duplication Analysis

**Test Code:** 1451

**Test Synonyms:**
- FANCA MLPA
- FA Complementation Group A
- Fanconi Anemia
- Fanconi Group A
- FANCA
- Deletion, Duplication

**CPT Code(s):** Incorrect (include sequencing)
- 83891
- 83900
- 83909 x 2
- 83914 x 42

**Background:**
Fanconi Anemia (FA) is characterized by bone marrow failure, increased risk for cancer, and physical abnormalities. Progressive bone marrow failure is responsible for the most significant morbidity and mortality. Clinically heterogeneous, FA individuals are at increased risk for acute myelogenous leukemia, myelodysplasia, and solid tumors of the neck, head, oral cavities, and gynecological system. Congenital abnormalities are present in approximately 70% of FA patients and include: café au lait spots or hypopigmentation; short stature; radial ray defects; eye defects such as microphthalmia; malformations of the kidney, genitalia, heart, gastrointestinal tract, ears, and feet. Currently, 15 genes have been identified that, when mutated, can cause FA. The Fanconi complementation group A gene, or FANCA, is inherited in an autosomal recessive manner. Mutations (small nucleotide variants as well as gross deletions/duplications) in FANCA are responsible for approximately 66% of all FA cases.

**Reasons for Referral:**
- Confirmation of clinical diagnosis in patients with classical or atypical FANCA.
- Carrier testing of family members of FANCA patients with known mutations.
- Prenatal diagnosis.

**Note:** Prior to submitting a sample for FANCA molecular testing, cytogenetic confirmation of Fanconi anemia, by breakage analysis, is recommended and can be performed in the Knight Diagnostic Laboratories.

For breakage analysis please contact Client Services at (855) 535-1522 to obtain the correct requisition form to accompany the specimen.

Additionally, prior to molecular testing, complementation typing (complementation group assignment) is strongly recommended. Complementation typing is available at a CLIA certified laboratory at the following URL address: [http://www.cincinnatichildrens.org/research/div/exphematology/translational/ttdsl.htm](http://www.cincinnatichildrens.org/research/div/exphematology/translational/ttdsl.htm).

Please attach the complementation group assignment documentation to the molecular testing requisition form, if possible.
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Methodology:
MLPA: Large deletions and duplications are detected using multiplex ligation-dependent probe amplification (MLPA).

Test reporting follows the ACMG Standards & Guidelines for Clinical Genetics Laboratories, Ultra-Rare Disorders Guidelines, and Interpretation of Sequence Variants Guidelines.

Specimen Requirements:
- **Blood**: Lavender tube (EDTA) or yellow (ACD)
  - Adult: 6.0 mL
  - Child: 6.0 mL
  - Infant: 2.0 mL

**A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES.** Please include detailed clinical information, including ethnicity, clinical history, and family history.

Test Performed (Days):
Weekly

Turn Around Time:
14 – 21 Days

Shipment Sensitivity Requirements:
Keep specimen cool during transit, but do not ship on dry ice. Please use the cold pack provided in the KDL shipping kit. Ship the specimen overnight express, using the FedEx priority overnight label provided. Contact Client Services at (855) 535-1522 for shipping kits and instructions.

Additional Information:
Related tests also performed in our laboratory: Full Sanger sequencing of FANCA gene.

Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact Client Services at (855) 535-1522.

References: