

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *PFIC1* Genetic Analysis
- **Test Code:** 3349

Related Diseases & Genes:

- "CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC, 1" (OMIM #: 211600)|| Gene: *ATP8B1* (OMIM #: 602397)

Synonyms:

-none-

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 5 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Multisystem Diseases

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For yesterday, today and *tomorrows...*

