

P53 CARRIER TESTING

Orderable – E-order/Requisition

Turnaround Time: 3 months

Alternate Name(s):

Li-Fraumeni Syndrome

Specimen:

Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

Interpretive Comments:

Germline mutations of the p53 gene are associated with Li-Fraumeni Syndrome, a rare autosomal dominant disorder characterized by a wide spectrum of tumours sarcomas, breast carcinomas, brain tumours and adrenocortical carcinomas. In most of the cases, tumours will develop in children and young adults. Germline p53 mutations are mostly missense mutations most commonly (>90%)**1,2** found within the DNA-binding domain of the p53 gene which is coded for by exons 5 through 8, and which can inactivate the transcriptional activity of the protein. These mutations can be identified by direct sequence analysis of PCR-amplified p53 gene coding sequence (exons) using genomic DNA derived from peripheral blood leukocytes as a template.

References

1. Frebourg T. "Germline mutations of the p53 gene" *Pathol Biol (Paris)*. 1997 Dec;45(10):845-51.
2. [p53 mutation database](#)



Laboratory:
Molecular Diagnostics Lab



Requisition:
[MOLECULAR DIAGNOSTIC REQUISITION](#)



Method of Analysis:
DNA Sequence Analysis



Test Schedule:
As required,
Monday to Friday 0800-
1600 hours



P53
CARRIER
TESTING

Critical Information Required:

Pedigree required

Storage and Shipment:

Must be received within 5 days of collection, shipped at room temperature by courier/overnight collection.