

# 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  
DIAGNOSTICS  
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.