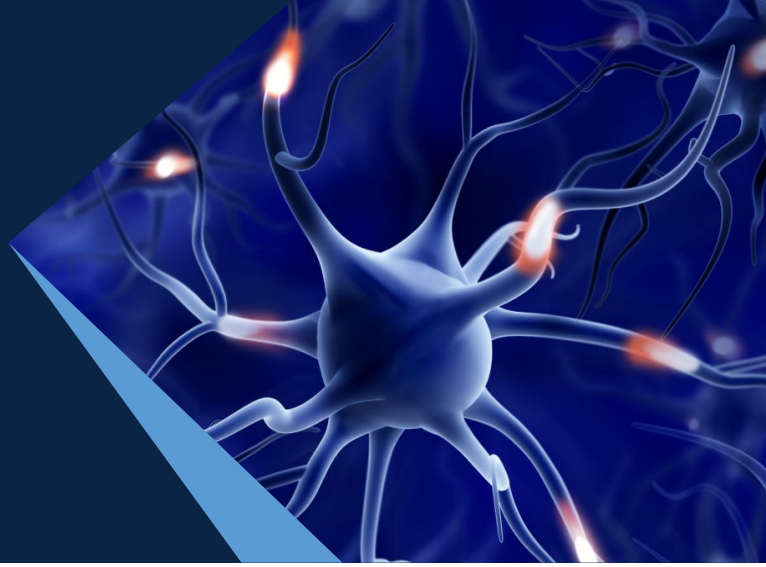
RML
MEHROTRA PATHOLOGY
Committed To Diagnostic Excellence

NEUROFIBROMATOSIS (NF)

Tumors Growing in the Nervous System



EFFICIENT DIAGNOSIS BY

MLPA -Multiplex ligation-dependent probe amplification

NEUROFIBROMATOSIS:

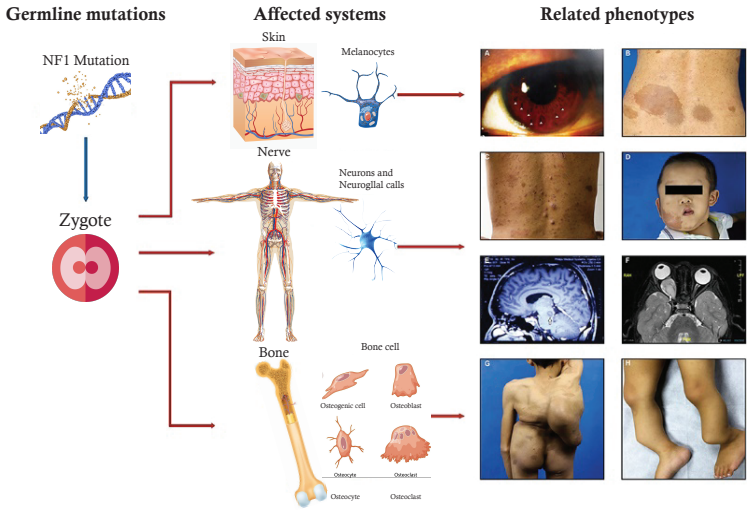
Autosomal dominant inherited disorder characterized by café-au-lait spots and fibromatous tumors of the skin. It develops tumors on nerve tissues including the brain, spinal cord and nerves that send signals between the brain and spinal cord and all other parts of the body.

NF-1 The NF1 gene, located on chromosome 17q11.2, produces the protein neurofibromin that helps regulate cell growth. The mutated gene causes a loss of neurofibromin, allowing cells to grow uncontrolled.

NF-2 The NF2 gene is located on chromosome 22q12.2, and produces a protein called merlin (also called schwannomin), which suppresses tumors. The mutated gene causes a loss of merlin, leading to uncontrolled cell growth.

Schwannomatosis- Caused by mutations of the tumor suppressor genes SMARCB1 and LZTR1.

Symptoms of NF-1 Neurofibromatosis



Responsible for Cancerous Tumors:

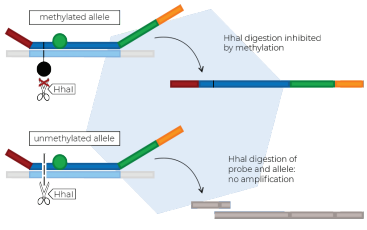
- Benign eye tumors/Lisch nodules/Glioma
- Brain tumors
- Adrenal gland tumors
- Muscle tumors
- Spinal cord tumors
- Malignant peripheral nerve sheath tumors (MPNST)/Sarcoma of nerve endings

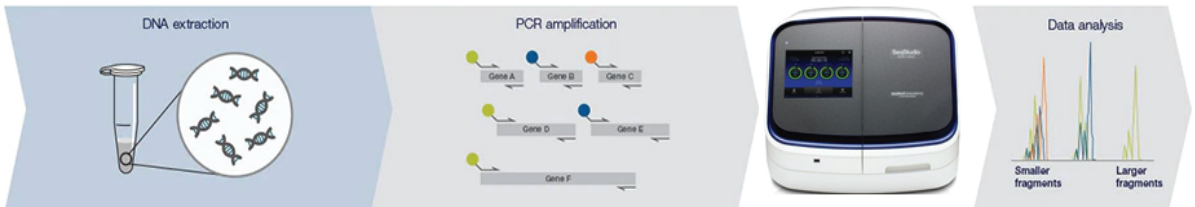
MLPA DIAGNOSIS ON SEQSTUDIO GENETIC ANALYZER

NF-1 MLPA detects deletions or duplications in the region surrounding the neurofibromin 1 (NF1) gene using 25 probes for the NF1 area on chromosome 17q11.2.

NF-2 MLPA detects deletions or duplications in the NF2 gene This includes 21 probes for the NF2 gene; 5 probes on chromosome 22q upstream of NF2, 4 of which target SMARCB1 and LZTR1; and 4 probes on chromosome 22q downstream of NF2

Schwannomatosis- MLPA detects deletions or duplications in the LZTR1 gene using 20 probes for the LZTR1 gene.





MLPA FRAGMENT ANALYSIS WORKFLOW

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