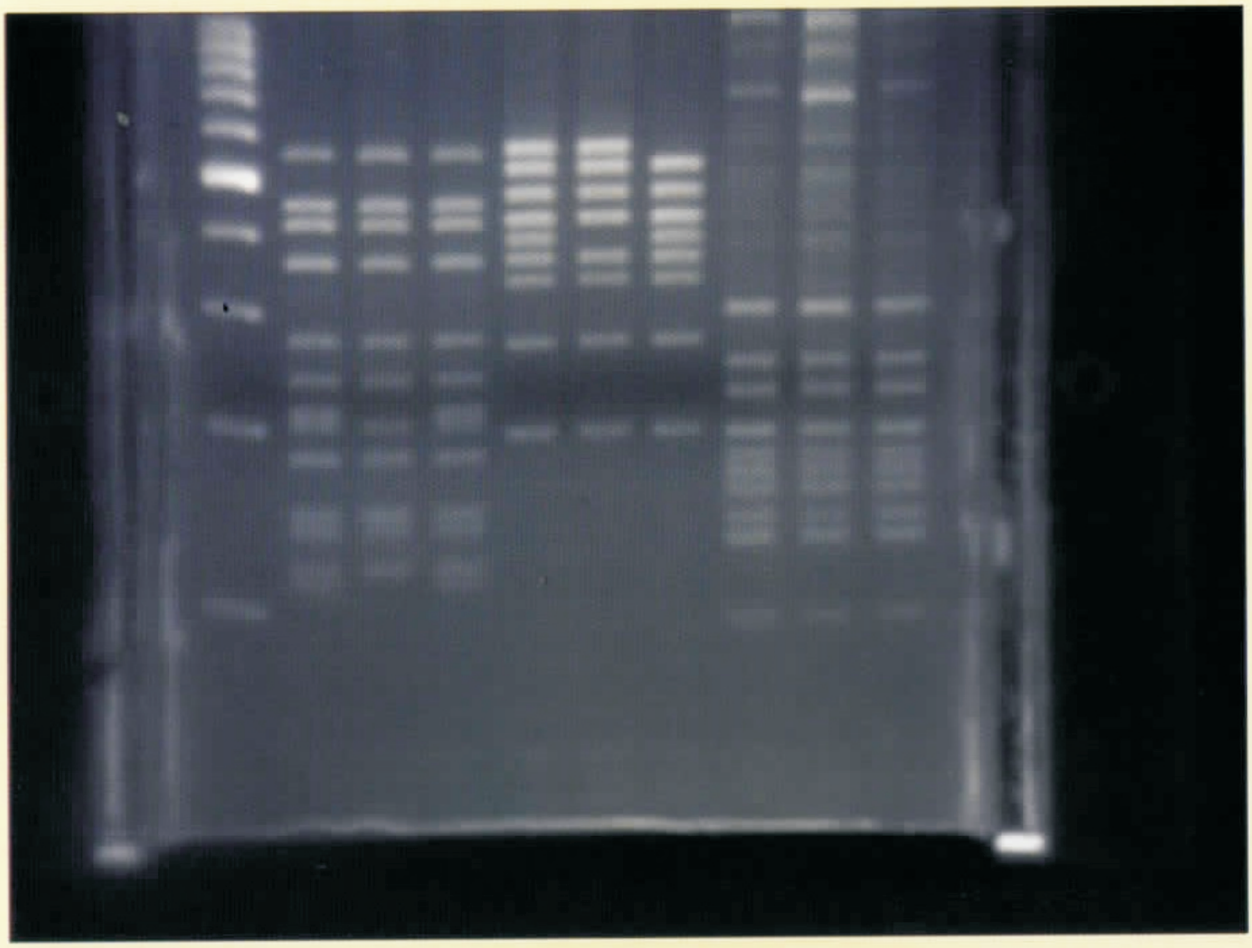
MLPA (MULTIPLE LIGATION DEPENDENT PROBE ANALYSIS) FOR DMD / BMD

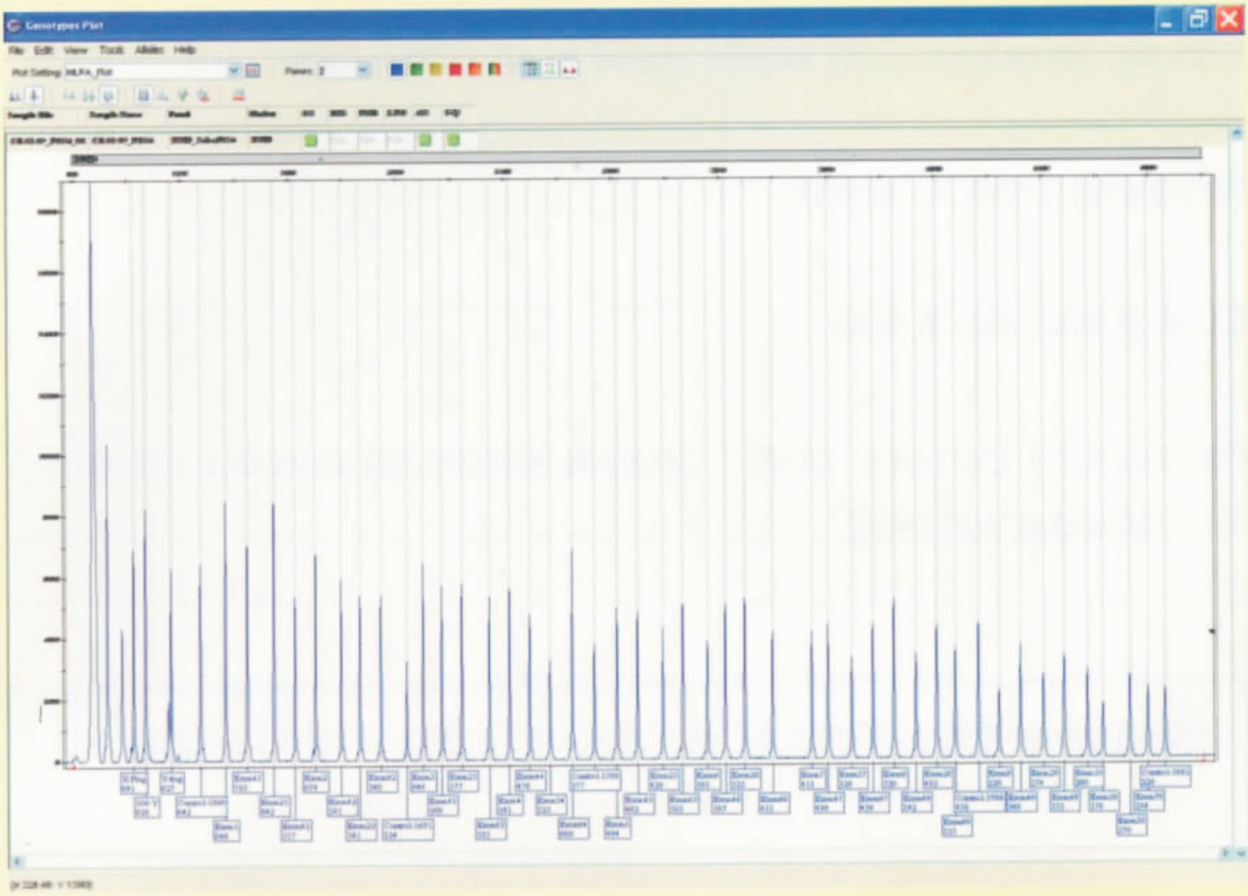
Genetic testing is often the best way to confirm a diagnosis in a patient with signs or symptoms suggestive of a genetic disease. Gene test also known as DNA based tests are performed on blood samples using sophisticated **PCR (Polymerase Chain Reaction)** techniques.

Testing using **DNA - based PCR technology** is now the **preferred diagnostic test** for one of the most common & severe Neuromuscular Disorders, namely **Duchenne Muscular Dystrophy (DMD)/Becker Muscular Dystrophy (BMD).** These diseases are caused by mutation i.e. changes, in different genes, hence are called genetic disorders.



Duchenne Muscular Dystrophy (DMD) is an X linked recessive disorder, and is the most common form of muscular dystrophy, occurring in about 1 in 3500 males. BMD is milder allelic form, with a lower prevalence. Typical clinical indications include elevated Creatine Kinase (CPK) levels, calf pseudohypertrophy, and difficulty in getting up from squatting position. DMD/BMD is caused due to mutations in DMD gene, which encodes the protein Dystrophin.

The **mPCR** (Multiplex Polymerase Chain Reaction) technique using **32** primer pairs offers a **rapid** simple and **affordable** screening method used to **detect** mutations **(deletions)** in **70% of male** patients. Approx 30% of other mutations including deletions (in other exons) and duplications are not detectable on mPCR analysis.



For those few patients who do not show deletions in the 32 exons screened by mPCR the advanced technique of **MLPA (Multiple Ligation dependent Probe Analysis)** for all **79 exons** involving PCR followed by capillary electrophoresis on Gene Sequencer can be carried out. This test helps to identify **deletions** in all 79 exons, **duplications**, which are found in around 10% of patients & **carrier status in females**. Point mutations are not detected by this method.

New MLPA Tests Offered:

1) Screening of all **79 exons** for **Deletions** & **Duplications.**
2) **Carrier** testing in **females** – If the mutations have been identified in the index case, carrier status can be determined in the affected patient's mother or adult sisters. **A written consent by the patient and the referring doctor is mandatory.**

Sample Required: 2 ml whole blood in an EDTA tube.

Reporting Time: 6 weeks

Cost of test: Rs. 8,500/-

Payment by Demand Draft / Cheque should be drawn in favour of "S.P. Mandali's IATRIS"

Test done from Monday to Friday (10.30am to 4.30 pm) at ;

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