# Genetic insights and counselling for a syndromic child presenting with Neurofibromatosis Type-1.

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## Background

This case study highlights the genetic counselling process for a family with 1-year-old male proband presenting with syndromic features, ultimately diagnosed with NF-1.

## Clinical data

The proband presented with poor weight gain and short stature. Physical examination revealed macrocephaly, hypotonia, cutis laxa, frontal bossing, flat feet, short stubby fingers, a pot belly, a systolic murmur, and developmental delay.

## Family history

The proband's parents are non-consanguineous, with no family history of consanguineous marriages. Initially, no similar congenital anomalies were reported among family members.

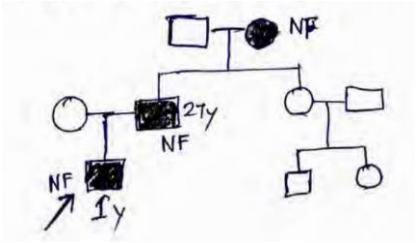
## Investigations

Given the multiple congenital anomalies, clinical exome sequencing was recommended by the treating physician.

## Results

The clinical exome sequencing revealed heterozygous, autosomal dominant mutations in the NF-1 gene.

## Pedigree chart




## Genetic counselling

### Pretest counselling:

Clinical exome sequencing was recommended due to the proband's multiple congenital anomalies. Details about testing centers, costs, and turnaround times were provided.

### Posttest counselling:

The findings of exome sequencing, its implications were explained, and information about NF-1 and its inheritance was shared. The proband's father and paternal grandmother showed similar NF-1 features. The risk of recurrence in future pregnancies and options for prenatal and preimplantation genetic screening were discussed. The proband was referred to Pediatrics for further follow-up and treatment.

## Conclusion

This case underscores the importance of genetic counselling in diagnosing and managing NF-1, providing crucial information and support for informed decision-making and ongoing management.