5 Things You Should Know ABOUT NEUROFIBROMATOSIS-1 (NF-1)

1 Genetic Basis and Inheritance

Neurofibromatosis Type 1 (NF1) is a genetic disorder caused by mutations in chromosome 17. NF1 follows an autosomal dominant inheritance pattern, meaning a child has a 50% chance of inheriting the condition if one parent has it.

2 Symptoms

- Café-au-lait spots:
- Neurofibromas
- Freckling in areas not typically exposed to the sun, such as the armpits or groin.
- Bone abnormalities like scoliosis (curvature of the spine) or tibial dysplasia (bowing of the legs)

3 Developmental Challenges

Approximately 50% of children with NF1 experience learning disabilities. These can include difficulties with attention, executive function, and visuospatial skills. Some children may also have speech and language delays.

4 Potential Complication

- Optic gliomas
- Skeletal abnormalities
- Cardiovascular issues
- Malignant peripheral nerve sheath tumors (MPNSTs):

5 Treatment

- Regular check-ups
- Surgical interventions
- Medications: Pain management and treatments for specific complications, such as optic gliomas.
- Supportive therapies: Including physical, occupational, and speech therapy to address developmental delays and cognitive issues