🧪 Rare Genetic Disorder Identified in Gujarat

📍 First Confirmed Case in India

Doctors at a tertiary care hospital in Ahmedabad have diagnosed India's first confirmed case of Bardet-Biedl Syndrome (BBS), a rare genetic disorder. The patient is a 9-year-old boy who presented with multiple systemic complications.

🧬 What is Bardet-Biedl Syndrome?

- **A rare inherited disorder caused by mutations affecting ciliary function.**
- **Belongs to the group of ciliopathies, which impact several organs.**
- **Global prevalence is estimated at 1 in 100,000–160,000 births, making it extremely rare.**

🔎 Clinical Presentation in the Child

The child was brought to the hospital with:

- **Obesity unresponsive to lifestyle changes.**
- **Progressive vision loss due to retinal degeneration.**
- **Polydactyly (extra fingers).**
- **Kidney abnormalities with declining function.**
- **Learning difficulties and developmental delays.**

🧪 Diagnosis & Genetic Confirmation

- **Initial clinical suspicion was based on the combination of obesity, vision impairment, and polydactyly.**
- **Whole Exome Sequencing confirmed pathogenic mutations consistent with Bardet-Biedl Syndrome.**
- **Multidisciplinary evaluation involving ophthalmology, nephrology, endocrinology, and genetics was carried out.**

💊 Treatment & Management

- **No curative treatment exists currently.**
- **Patient is being managed with:**
    - **Low-vision aids and ophthalmic follow-up.**
    - **Dietary interventions to control obesity.**
    - **Nephrology support for kidney monitoring.**
    - **Developmental therapy for learning challenges.**
- **Family provided with genetic counseling to understand inheritance patterns and future planning.**

🌍 Significance for India

- **The first confirmed case highlights the importance of genetic testing in rare diseases.**
- **Opens opportunities for research collaborations in India on ciliopathies.**
- **The hospital plans to maintain a registry of rare genetic disorders for better understanding and management.**