DECCAN Chronicle

Fragile X Syndrome in children

Doctors urge parents to watch for developmental delays, social withdrawal, and hyperactivity — potential red flags of this genetic disorder

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When three-year-old Varun began struggling in playschool, his parents grew increasingly concerned. His cognitive development lagged, and he showed little interest in learning. Ten-year-old Eshani, meanwhile, withdrew socially from a young age, barely engaging with friends or family. Teenager Ishaan faced attention deficits, emotional outbursts, and struggled to adapt to change.

Though different in age and symptom, all three children may be grappling with the same underlying condition — Fragile X Syndrome (FXS), a genetic disorder that often goes undetected until developmental delays become too evident to ignore.

KNOW THE RED FLAGS

- Speech or language delays
- Learning disabilities
- Repetitive speech or movements
- Poor social interaction
- Sensory sensitivities (light, noise, textures)
- Difficulty with transitions
- Anxiety or mood swings
- Autism-like traits

If your child displays several of these signs, consult a developmental specialist. Early action can change the course.

WHAT IS FRAGILE X SYNDROME?

Dr Suhas, Vice President of Bangalore Hospital, explains that FXS is a hereditary disorder caused by a mutation in the FMR1 gene located on the X chromosome. "This mutation disrupts the production of a protein (FMRP) essential for brain development," he says.

"IN SPECIFIC CASES, NEUROIMAGING LIKE 3T MRI CAN AID IN UNDERSTANDING BRAIN FUNCTION AND DRAFTING PERSONALISED THERAPEUTIC PLANS. A HOLISTIC, COORDINATED APPROACH BETWEEN HEALTHCARE, SCHOOL AND FAMILY SUPPORT SYSTEMS IS VITAL FOR LONG-TERM IMPROVEMENT." — DR MALINI SABA

'THE BRAIN IS A DELICATE MACHINE'

Psychoanalyst **Dr Malini Saba**, founder of the Saba Family Foundation, adds: "FXS can affect how a person learns and responds to the world. A small shift in DNA can alter how we react emotionally. It's a reminder of how complex the brain is."

EARLY SIGNS TO WATCH FOR CHILDREN WITH FRAGILE X MAY DISPLAY:

- Speech delays
- Attention issues
- Hyperactivity
- Emotional meltdowns
- Sleep disturbances
- Learning disabilities
- Autism-like behaviours (e.g., hand-flapping, avoiding eye contact)
- Difficulty adapting to routine changes

Physical signs are subtle in early stages, but neuroimaging like 3T MRI can offer insight into underlying brain changes.

NO CURE, BUT HELP IS AVAILABLE

"While there's no cure for FXS, symptom management through early intervention makes a significant difference," says Dr Suhas.

In some cases, medication is prescribed for anxiety, mood instability, or seizures.

Dr Saba adds, "There's no antidote, but getting timely help really changes outcomes. Therapies must be tailored to each child's needs — helping with emotional regulation, communication, motor skills, and daily functioning."

WHY BOYS ARE MORE AFFECTED

FXS affects boys more severely than girls. "Males often face more pronounced intellectual and behavioural challenges, while females may experience subtler learning or emotional difficulties," notes Dr Suhas.

THE GENETICS BEHIND IT

FXS stems from CGG repeats — a DNA sequence within the FMR1 gene. If the number of repeats exceeds 200, the gene becomes inactive, halting FMRP production. This mutation can be passed down through generations. Women can be silent carriers, showing no symptoms but passing on the mutation to their children.

HOPE LIES IN RESEARCH

Though current science can't reverse the mutation, researchers are exploring gene therapy and protein replacement. "We're not there yet," admits Dr Suhas, "but science is advancing."

In the meantime, families are urged to recognise the signs early, seek support, and advocate for coordinated care across schools, homes, and healthcare systems.