Syndromic Craniosynostosis Diagnosis

Diagnosing syndromic craniosynostosis is a multidisciplinary process that integrates clinical assessment, radiological findings, and genetic testing. Early and accurate diagnosis is crucial for guiding management and anticipating complications.

Clinical Evaluation

A detailed clinical history and physical examination are the foundation of diagnosis. Key clinical features include:

Multisuture craniosynostosis – as opposed to isolated sagittal or coronal fusion

Craniofacial anomalies – such as midface hypoplasia, orbital hypertelorism, or beaked nose

Limb anomalies – syndactyly (especially in Apert Syndrome)

Airway abnormalities – snoring, apneas, stridor

Developmental delay – variable depending on the syndrome

Family history – may suggest autosomal dominant inheritance

Radiological Assessment

Imaging plays a central role in confirming suture fusion and identifying associated malformations:

CT scan with 3D reconstruction – gold standard for evaluating suture fusion patterns and skull morphology

MRI – assesses brain development and associated conditions like Chiari Malformation, Hydrocephalus, or Skull Base Anomaly

Lateral skull X-ray – may provide initial clues in neonates

Key features to assess include:

Shape of the cranial vault

Patency of cranial sutures

Volume of posterior fossa

Presence of Platybasia, Basilar Invagination, or other Craniovertebral Junction anomalies

Genetic Testing

Molecular confirmation is essential to distinguish between syndromic and non-syndromic forms:

Targeted gene panels or whole-exome sequencing

Common mutations involve FGFR1, FGFR2, FGFR3, TWIST1, and others

Syndromic forms often have known gene associations:

Crouzon Syndrome → FGFR2

Pfeiffer Syndrome → FGFR1 / FGFR2

Apert Syndrome → FGFR2

Saethre Chotzen Syndrome → TWIST1

Genetic counseling is recommended for affected families.

Multidisciplinary Team Involvement

Diagnosis and follow-up should involve:

Neurosurgery

Craniofacial surgery

Clinical genetics

Pediatrics

Otolaryngology, Ophthalmology, and Pulmonology as needed