Skull Base Anomaly

A skull base anomaly refers to a congenital or acquired malformation affecting the bones and structures at the base of the skull, often impacting the craniocervical junction and nearby neurovascular elements. These anomalies can alter the normal anatomy and alignment between the brain, brainstem, and upper cervical spine, sometimes resulting in compression or instability.

Common types of skull base anomalies include:

Platybasia – abnormal flattening of the skull base angle

Basilar Invagination – upward displacement of the odontoid into the foramen magnum

Odontoid Retroflexion – posterior angulation of the odontoid process toward the brainstem

Chiari Malformation – descent of cerebellar tonsils below the foramen magnum, often associated

Craniosynostosis – premature fusion of cranial sutures, which may secondarily affect the skull base

Craniovertebral Junction anomalies – developmental defects of the occiput, atlas, or axis

These anomalies are particularly relevant in patients with Syndromic Craniosynostosis, where early fusion of skull base synchondroses can predispose to such malformations.

Clinical Relevance

Symptoms may include:

Headaches

Brainstem compression signs

Myelopathy

Hydrocephalus

Sleep apnea

Diagnosis is typically made through midsagittal MRI and CT imaging, using specific angles and morphometric parameters like the basal angle, clivoaxial angle, and pB-C2 distance.

Management depends on the severity and associated symptoms, ranging from observation to Neurosurgical decompression and Craniovertebral Fusion.

Retrospective multicenter studies

In a retrospective multicenter study by Pablo M. Munarriz and collaborators, they address the underexplored area of cranial base anomaly and craniocervical junction malformations in patients

with genetically confirmed syndromic craniosynostosis [1].

The study offers valuable insights into the prevalence and progression of abnormalities such as [Chiari malformation type I] (CMI), platybasia, odontoid retroflexion, basilar invagination, and syringomyelia.

A major strength lies in the systematic radiological evaluation using midsagittal MRI metrics, including basal angle, pB-C2, and clivoaxial angle, providing objective and reproducible anatomical criteria. The inclusion of a follow-up MRI in 56 out of 77 patients adds a dynamic perspective to disease evolution.

The study reveals a statistically significant increase in CMI frequency over time (from 7.8% to 21.4%, P = .021), especially within Crouzon syndrome and Pfeiffer syndrome patients. This finding supports the hypothesis of progressive cerebellar tonsillar descent associated with cranial base growth abnormalities and potentially increased intracranial pressure. Conversely, other malformations such as basilar invagination and odontoid retroflexion remained rare, with only marginal changes over time. Interestingly, platybasia declined in prevalence, possibly indicating developmental compensation or variability in diagnostic criteria.

However, some limitations should be noted. The study does not explore clinical correlations (e.g., headache, myelopathy), limiting the translational relevance of the radiological findings. Furthermore, although the cohort is relatively large for a rare disease study, the subgroup analysis by syndrome is underpowered for definitive comparisons. Longitudinal follow-up periods and potential confounders (e.g., prior surgeries) are not detailed.

In summary, this paper enriches our understanding of the natural history of cranial base anomalies in syndromic craniosynostosis, highlighting the need for vigilant long-term radiological monitoring in selected syndromes. Future studies should aim to integrate clinical outcomes and investigate the impact of early surgical intervention on the evolution of CMI and related pathologies.

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Munarriz PM, Rius-Diaz F, Panero I, Martinez de Aragon A, López-Bermeo D, Saceda J, Rivero B, Miranda-Lloret P, Iglesias S. Skull Base and Craniocervical Anomalies in Syndromic Craniosynostosis. Neurosurgery. 2025 Apr 11. doi: 10.1227/neu.0000000000003433. Epub ahead of print. PMID: 40214267.