

DIAGNOSTIC ACCURACY OF ULTRASOUND IN DIAGNOSING CRANIOSYNOSTOSIS IN INFANTS USING CT AS THE GOLD STANDARD

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ABSTRACT

OBJECTIVE

This study aimed to determine the diagnostic accuracy of cranial ultrasound in detecting craniosynostosis in infants using computed tomography (CT) as the reference standard.

METHODOLOGY

This cross-sectional validation study was conducted in the Department of Radiology, MTI-Hayatabad Medical Complex, Peshawar. A total of 125 infants aged 2–6 months with clinical suspicion of craniosynostosis were enrolled through non-probability consecutive sampling. All infants underwent cranial ultrasound followed by CT scan. Ultrasound findings were compared with CT results, which were taken as the gold standard. Sensitivity, specificity, positive predictive value (PPV), negative predictive value (NPV), and overall diagnostic accuracy were calculated.

RESULTS

The mean age of infants was 4.02 ± 1.40 months. CT confirmed craniosynostosis in 120 (96.0%) cases. Ultrasound demonstrated a sensitivity of 79.17% (95% CI: 71.05–85.47%), specificity of 80.00% (95% CI: 37.55–96.38%), PPV of 98.96% (95% CI: 94.33–99.82%), NPV of 13.79% (95% CI: 5.50–30.56%), and overall diagnostic accuracy of 79.20% (95% CI: 71.27–85.39%).

CONCLUSION

Cranial ultrasound shows acceptable diagnostic accuracy with very high PPV, supporting its use as a useful adjunct first-line imaging tool for craniosynostosis in settings with a high pre-test probability of disease, particularly in resource-limited settings; however, its low NPV in this high-prevalence referral cohort means a negative ultrasound cannot reliably exclude the diagnosis, and CT remains essential for equivocal cases and surgical planning.

KEYWORDS: Craniosynostosis; Cranial Ultrasound; Computed Tomography; Diagnostic Accuracy; Infant Imaging; Radiation-Free Screening; Pediatric Radiology

INTRODUCTION

Craniosynostosis is a congenital craniofacial anomaly characterised by premature fusion of one or more cranial sutures, resulting in abnormal skull morphology and potential restriction of normal brain growth.¹ If not identified and treated early, craniosynostosis may lead to raised intracranial pressure, neurodevelopmental delay, visual impairment, and psychosocial consequences. Early diagnosis is therefore critical to facilitate timely surgical intervention and optimise neurodevelopmental outcomes.² Recent epidemiological data estimate the global incidence of craniosynostosis to be approximately 1 in 2,000–2,500 live births, with an apparent rise in reported cases attributed to improved diagnostic awareness and imaging availability.³ Craniosynostosis can occur as an isolated, non-syndromic condition or as part of a genetic syndrome. Non-syndromic craniosynostosis

accounts for nearly 75% of cases, commonly involving sagittal, coronal, metopic, or lambdoid sutures. Genetic mutations, often involving fibroblast growth factor receptor pathways, contribute to approximately 20% of cases, many arising de novo. Environmental factors such as intrauterine constraint, maternal smoking, and teratogen exposure have also been implicated. Regardless of aetiology, early radiological confirmation is essential once clinical suspicion arises.⁴ Computed tomography (CT) with three-dimensional reconstruction remains the gold standard imaging modality for diagnosing craniosynostosis due to its superior spatial resolution and detailed depiction of cranial sutures. CT also plays a vital role in surgical planning.⁵ However, CT exposes infants to ionising radiation, which has been associated with an increased lifetime risk of malignancy. Contemporary pediatric imaging guidelines emphasise minimising radiation exposure and adhering to the ALARA (As Low As Reasonably

Achievable) principle, particularly in early infancy Cranial ultrasound has emerged as an attractive alternative imaging modality in this context.⁶ It is non-invasive, radiation-free, widely available, and cost-effective. The presence of open fontanelles in early infancy allows high-frequency linear probes to visualise cranial sutures directly. Normal sutures appear as hypoechoic gaps between hyperechoic bony margins, whereas fused sutures demonstrate narrowing, bridging, or complete absence of this gap.⁷ Over the past five years, several studies have reported promising diagnostic performance of ultrasound, particularly for sagittal and coronal sutures, when performed by trained radiologists using standardised protocols.⁸ In low- and middle-income countries such as Pakistan, the burden of unnecessary CT scans is particularly significant. Limited access to advanced imaging, financial constraints, and lack of paediatric anaesthesia services further complicate timely diagnosis.⁹ Establishing ultrasound as a reliable first-line screening tool could substantially reduce radiation exposure, healthcare costs, and diagnostic delays. Despite growing international evidence, local data on the diagnostic accuracy of cranial ultrasound remain limited. This study was therefore designed to evaluate the diagnostic accuracy of cranial ultrasound in detecting craniosynostosis in infants using CT scan as the reference standard. By providing local evidence from a tertiary care centre, this study aims to support safer, more accessible diagnostic pathways and inform imaging practices in resource-constrained settings.

METHODOLOGY

This cross-sectional validation study was conducted at the Department of Radiology, MTI-Hayatabad Medical Complex, Peshawar, after approval from the Hospital Ethical Review Board (Approval No. 1725) and the Research Evaluation Unit, CPSP Karachi. Written informed consent was obtained from parents or legal guardians. Infants aged 2-6 months referred with clinical suspicion of craniosynostosis were enrolled through non-probability consecutive sampling. Infants outside the specified age range, those without consent, and those who did not undergo both ultrasound and CT during the same diagnostic work-up were excluded. As recruitment was limited to clinically suspected cases at a tertiary referral centre, the study population represented a high-pretest-probability cohort. Cranial ultrasound was performed before CT using a high-frequency 14 MHz linear transducer. Major cranial sutures were systematically examined in accessible longitudinal and transverse planes. Patent sutures were identified as hypoechoic gaps between echogenic bony

margins, while narrowing, loss of the normal gap, or osseous bridging was considered suggestive of premature fusion. Ultrasound was considered positive when fusion of at least one cranial suture was detected. CT of the skull served as the reference standard, with suture patency assessed on bone-window images and multiplanar or three-dimensional reconstructions where available. Ultrasound interpretation preceded CT; however, formal blinding of CT readers to ultrasound findings was not prospectively documented. All 125 infants with paired ultrasound and CT results were analysed. A formal a priori sample-size calculation was not documented. The 120 CT-positive cases provided adequate precision for estimating sensitivity, whereas the small number of CT-negative cases limited the precision of specificity and negative predictive value. Sensitivity, specificity, positive and negative predictive values, overall diagnostic accuracy, and disease prevalence were calculated from a 2×2 table with 95% exact binomial confidence intervals. Data were analysed using SPSS version 23. Suture-specific diagnostic accuracy was not calculated because complete paired suture-level ultrasound and CT data were unavailable.

RESULTS

A total of 125 infants were included. The mean age was 4.02 ± 1.40 months, with 61 (48.8%) males and 64 (51.2%) females (Table I). CT confirmed craniosynostosis in 120 (96.0%) infants (Table II). Ultrasound correctly identified 95 true-positive and 4 true-negative cases. There were 25 false-negative and 1 false-positive cases (Table III). Sensitivity, specificity, PPV, NPV, and overall diagnostic accuracy were 79.17%, 80.00%, 98.96%, 13.79%, and 79.20%, respectively (Table IV).

Table 1: Demographic Characteristics of Study Population (n=125)

Variable	Value
Mean age(months)	4.02 ± 1.40
Gender (Male/Female)	61 (48.8%) / 64 (51.2%)
Total Infants	125

Table 2: CT-Confirmed Craniosynostosis (n=125)

CT Finding	Frequency (%age)
Craniosynostosis Present	120 (96.0%; 95% CI 90.91-98.69)
Absent	5 (4.0%)

Table 3: Ultrasound Vs CT Diagnostic Comparison

	CT Positive	CT Negative
US Positive	95	1
US Negative	25	4

Table 4: Diagnostic Performance of Ultrasound

Parameter	Estimate	95% age CI
Sensitivity	79.17%	70.80-86.04
Specificity	80.00%	28.36-99.49
PPV	98.96%	94.33-99.97
NPV	13.79%	3.89-31.66
Overall Accuracy	79.20%	71.03-85.94

DISCUSSION

The present study demonstrates that cranial ultrasound has acceptable diagnostic accuracy with a very high positive predictive value for detecting craniosynostosis in infants when compared with CT scan. These findings support the growing body of evidence advocating ultrasound as a useful adjunct first-line imaging tool in early infancy for infants with a high pre-test probability of craniosynostosis; however, given the low NPV observed in this high-prevalence cohort, a negative ultrasound finding alone should not be considered sufficient to exclude the diagnosis when clinical suspicion persists, and such cases should proceed to CT. The sensitivity and specificity observed in this study are consistent with recent international literature.¹⁰ Martino et al. (2022) reported sensitivity exceeding 80% for ultrasound in detecting sagittal and coronal suture fusion, while Najmi et al. (2023) demonstrated comparable diagnostic accuracy in a multicenter cohort. High PPV in our study indicates that a positive ultrasound finding strongly correlates with true craniosynostosis, allowing clinicians to proceed with referral and management.^{11,12} The low negative predictive value observed in this study warrants careful interpretation. This finding is primarily attributable to the very high prevalence of CT-confirmed craniosynostosis in our cohort, reflecting referral bias inherent to a tertiary care setting. Negative predictive value is known to be highly influenced by disease prevalence rather than intrinsic test performance. The 96% prevalence of CT-confirmed craniosynostosis observed in this cohort is markedly higher than the estimated population prevalence of approximately 1 in 2,000-2,500 live births, and reflects the fact that infants were drawn exclusively from a tertiary-care radiology referral pathway in which clinical pre-selection for craniosynostosis was already high, rather than from an unselected or primary-care population.³ By Bayes' theorem, NPV falls as pre-test prevalence rises, even when the underlying sensitivity and specificity of a test remain constant; the very low NPV reported here is therefore a predictable consequence of this referral-driven prevalence rather than evidence of poor intrinsic test performance. Consequently, these findings are most directly applicable to tertiary-referral populations with a similarly high pre-test probability of disease, and

should not be extrapolated to lower-prevalence settings such as primary-care or population-based screening, where the same sensitivity and specificity would be expected to yield a substantially higher NPV. Similar trends have been reported in high-prevalence referral populations.¹³ False-negative ultrasound cases in our study were mainly associated with partial suture fusion and posterior suture involvement, particularly lambdoid sutures. These limitations have been consistently documented in prior studies and highlight the importance of operator expertise and systematic scanning protocols.¹⁴ Suture-specific diagnostic performance (i.e., separate sensitivity and specificity for the sagittal, coronal, metopic, and lambdoid sutures) is not reported in the present analysis. Consequently, CT remains indispensable in cases with negative or equivocal ultrasound findings when clinical suspicion persists, as well as for detailed preoperative planning.¹⁵ Future multicenter studies should recruit a broader spectrum of referred and non-referred infants, pre-specify sample-size targets for both sensitivity and specificity, use standardised ultrasound acquisition and interpretation criteria, document reader experience and blinding, and report suture-specific performance. Such studies would better define where ultrasound can safely reduce CT use without delaying diagnosis.

CONCLUSIONS

Cranial ultrasound showed moderate sensitivity and a high positive predictive value for craniosynostosis in this highly selected tertiary-referral cohort. However, the low negative predictive value and the wide confidence interval around specificity mean that ultrasound should not be used as a stand-alone rule-out test. It may be used as a radiation-free initial adjunct or triage examination in selected clinically suspected infants, while CT remains the reference standard when ultrasound is negative or equivocal despite persistent clinical suspicion and for surgical planning.

LIMITATIONS

The single-centre tertiary-referral design, extreme disease prevalence, the very small CT-negative subgroup, and the resulting imprecision of specificity and NPV. Operator experience was not stratified analytically, CT-reader blinding to ultrasound was not prospectively documented, and suture-specific paired accuracy could not be assessed. Although the number of CT-positive cases provides reasonable precision for the sensitivity estimate, the study is underpowered for a precise estimate of specificity.

CONFLICT OF INTEREST: None

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REFERENCES

- Bautista G. Craniosynostosis: neonatal perspectives. *Neoreviews*. 2021;22(4). doi:10.1542/neo.22-4-e250. PMID: 33795400.
- Bashawieh OO, Alyami MH, Alghamdi MA, Alotaibi FE, Alluhaybi AA, Altuhaini KS. Neurodevelopmental outcomes in children with craniosynostosis: a retrospective cross-sectional analysis. *Childs Nerv Syst*. 2026;42(1):49. doi:10.1007/s00381-026-07128-9. PMID: 41604010.
- Lewanda AF, Jabs EW, Sanchez-Lara PA. Craniosynostosis. In: Pyeritz RE, Korf BR, Grody WW, editors. *Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Developmental Disorders*. Elsevier; 2025. p. 81-104. doi:10.1016/B978-0-12-815234-8.00004-5. PMID: 31335086.
- Lattanzi W. Craniosynostosis: genetic basis, genes, chromosomes, and resulting syndromes. In: Di Rocco C, Pang D, Rutka JT, editors. *Textbook of Pediatric Neurosurgery*. Cham: Springer; 2020. p. 1373-1391. doi:10.1007/978-3-319-72168-2_56.
- Russo C, Aliberti F, Ferrara UP, Russo C, De Gennaro DV, Cristofano A, et al. Neuroimaging in non-syndromic craniosynostosis: key concepts to unlock innovation. *Diagnostics (Basel)*. 2024;14(17):1842. doi:10.3390/diagnostics14171842.
- Aziz MM, Onyejesi C, Pyala R, Alattar O, Abdul AA, Alagarswamy K, et al. Reducing radiation exposure in pediatric CT imaging: strategies and alternatives in emergency medicine—a narrative review. *J Emerg Crit Care Med*. 2025;9. doi:10.21037/jcccm-24-102.
- Caro-Domínguez P, Lecacheux C, Hernandez-Herrera C, Llorens-Salvador R. Cranial ultrasound for beginners. *Transl Pediatr*. 2021;10(4):1117-1137. doi:10.21037/tp-20-399. PMID: 34012859.
- Micovic M, Zivkovic B, Vukasinovic I, Jelovac D, Stojicic M, Bascarevic V. Imaging modalities in craniosynostosis: a systematic review and proposal of the ARCANA protocol for multimodal radiation-free assessment. *Diagnostics (Basel)*. 2025;15(20):2632. doi:10.3390/diagnostics15202632.
- Massimi L, Bianchi F, Frassanito P, Calandrelli R, Tamburrini G, Caldarelli M. Imaging in craniosynostosis: when and what? *Childs Nerv Syst*. 2019;35(11):2055-2069. doi:10.1007/s00381-019-04278-x. PMID: 31289853.
- Proisy M, Bruneau B, Riffaud L. How ultrasonography can contribute to diagnosis of craniosynostosis. *Neurochirurgie*. 2019;65(5):228-231. doi:10.1016/j.neuchi.2019.09.019. PMID: 31586456.
- Marino S, Ruggieri M, Marino L, Falsaperla R. Sutures ultrasound: useful diagnostic screening for posterior plagiocephaly. *Childs Nerv Syst*. 2021;37(12):3715-3720. doi:10.1007/s00381-021-05324-3. PMID: 34453581.
- Alizadeh H, Najmi N, Mehdizade M, Najmi N. Diagnostic accuracy of ultrasonic examination in suspected craniosynostosis among infants. *Indian Pediatr*. 2013;50(1):148-150. doi:10.1007/s13312-013-0029-9. PMID: 23396787.
- Montoya JC, Eckel LJ, DeLone DR, Kotsenas AL, Diehn FE, Yu L, et al. Low-dose CT for craniosynostosis: preserving diagnostic benefit with substantial radiation dose reduction. *AJNR Am J Neuroradiol*. 2017;38(4):672-677. doi:10.3174/ajnr.A5063. PMID: 28183836.
- Lyoo Y, Choi YH, Lee SB, Lee S, Cho YJ, Shin SM, et al. Ultra-low-dose computed tomography with deep learning reconstruction for craniosynostosis at radiation doses comparable to skull radiographs: a pilot study. *Pediatr Radiol*. 2023;53(11):2260-2268. doi:10.1007/s00247-023-05717-3. PMID: 37488451.
- Kaasalainen T, Männistö V, Mäkelä T, Suojanen J, Nuorala A, Heliövaara A, et al. Postoperative computed tomography imaging of pediatric patients with craniosynostosis: radiation dose and image quality comparison between multi-slice computed tomography and O-arm cone-beam computed tomography. *Pediatr Radiol*. 2023;53(8):1704-1712. doi:10.1007/s00247-023-05644-3. PMID: 36967418.
- Tao W, Goetti R. Evaluation of ultra-low-dose CT with tin filter for craniosynostosis. *J Med Imaging Radiat Oncol*. 2025;69(1):28-34. doi:10.1111/1754-9485.13812. PMID: 39601235.
- de Goederen R, Yang S, Pullens B, Wolvius EB, Joosten KFM, Mathijssen IMJ. Evaluation of the OSA treatment protocol in syndromic craniosynostosis during the first 6 years of life. *J Plast Reconstr Aesthet Surg*. 2021;74(10):2674-2682. doi:10.1016/j.bjps.2021.03.033. PMID: 34112566.
- Fahradyan A, Daneshgaran G, Hoffman TL, Wexler A, Francis SH. Challenging the norm: is routine use of cranial CT in evaluation of craniosynostosis necessary? *J Craniofac Surg*. 2021;32(7):2496-2499. doi:10.1097/SCS.00000000000007926. PMID: 34705393.

AUTHORS CONTRIBUTION

Kiran Zafar - Concept & Design; Data Acquisition; Data Analysis/Interpretation; Drafting Manuscript; Critical Revision; Supervision; Final Approval
Naila Tamkeen - Concept & Design; Data Acquisition; Data Analysis/Interpretation; Drafting Manuscript; Critical Revision; Supervision; Final Approval
Jawad Ali - Concept & Design; Data Acquisition; Data Analysis/Interpretation; Drafting Manuscript; Critical Revision; Supervision; Final Approval
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Sahar Gulab - Concept & Design; Data Acquisition; Data Analysis/Interpretation; Drafting Manuscript; Critical Revision; Supervision; Final Approval
Faryal Bashir - Concept & Design; Data Acquisition; Data Analysis/Interpretation; Drafting Manuscript; Critical Revision; Supervision; Final Approval
The authors accept responsibility for all aspects of the work and will ensure that any concerns regarding the accuracy or integrity of any part are properly investigated and resolved.



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