Oregon Health & Science University
School of Medicine

Scholarly Projects Final Report

Title

Craniosynostosis: A Radiographic Review

Student Investigator's Name

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Date of Submission

3/11/2024

Graduation Year

2024

Project Course

Scholarly Project

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OHSU Diagnostic Radiology

Scholarly Project Final Report

Concentration Lead's Name

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Project/Research Question

How can the various craniosynostosis subtypes and their associated clinical outcomes be best characterized from a modern, radiographic perspective?

Project aims:
1) Describe the radiographic features of craniosynostosis
2) Understand the calvarial deformities associated with untreated craniosynostosis
3) Differentiate between the available treatment options for craniosynostosis
4) Review expected post-operative imaging findings after cranial vault reconstruction
5) Identify similar presenting conditions that warrant diagnostic consideration

Type of Project

Research Project

Keywords

Radiology, Pediatrics, Craniosynostosis, Skull, Congenital

Meeting Presentations

OHSU Research Week 2023, Portland, Oregon, May 2, 2023, Podium Presentation

American Society of Head & Neck Radiology 2023 Annual Meeting, Orlando, Florida, September 20, 2023, Educational Exhibit

Submission to Archive

None to date

Next Steps

The database could be used if a colleague desires to pursue a more specific craniosynostosis-related project.

Scholarly Project Final Report

Introduction

Craniosynostosis is a common pediatric condition defined by the premature fusion of cranial sutures and is known to cause visually distinct cranial abnormalities[1]. While each cranial suture has a typical timeline for its physiologic closure[2], none should begin to fuse before the age of about two years (except the metopic suture, which closes during the first year of life). It is difficult to assess for individual premature sutural closure without the use of imaging, and therefore calvarial deformities remain the most recognizable sign of potential underlying craniosynostosis. For example, sagittal synostosis with associated scaphocephaly is the most observed subtype of craniosynostosis, in which premature fusion of the sagittal suture causes the head to grow in an abnormally long, narrow pattern (see Figure 3).

Because the use of imaging (particularly CT with 3D reconstruction) is integral to the accurate diagnosis of craniosynostosis, the continued refinement of how this condition is radiographically characterized is important for the continued advancement of the treatment of this condition. The diagnosis of craniosynostosis begins with the clinical assessment and ends with radiographic diagnosis and is time-sensitive. Prolonged, untreated craniosynostosis can cause a permanently misshapen skull with associated self-esteem and cosmetic insecurity, as well as complications related to increased cranial pressure and compression of internal CNS structures[3]. The purpose of this study is to outline the most up-to-date radiographic considerations when any physician (whether this is the pediatrician, neurologist/neurosurgeon, or radiologist) is attempting to diagnose craniosynostosis.

While the radiologist is the physician most specifically trained in diagnosing conditions through medical image analysis, this imaging is a resource read and used by most practicing physicians. When considering the time-sensitive nature of craniosynostosis, it is important to equip the entire population of physicians with the knowledge necessary to make informed decisions about pediatric medical care, especially when a radiology report is unavailable. Additionally, the evolving COVID-19 pandemic has shown what the extreme of limited healthcare access in the US can look like. This includes scheduling delays for all imaging modalities, financial strains on patients and institutions, and the need to triage and prioritize diagnostic workups from patient to patient[4]. Alongside these system-wide shifts, the quality and technology behind medical imaging itself have modernized radically in the time that craniosynostosis was first described in medical literature in the early 1800s[5]. Therefore, this study will provide a comprehensive review of the radiographic features and relevant considerations of craniosynostosis so that any physician can adequately advise on potential cranial abnormalities in the pediatric population.

Methods

Study Design

This retrospective, single-institutional study was performed through chart review at Oregon Health & Science University. To quickly search, filter, and access radiology reports of interest to the below population, an internal search engine called “RadSearch” was utilized[6]. RadSearch allowed for streamlined patient searching, access to relevant radiology reports while restricting access to unneeded PHI, and

hyperlink integration to pull up the associated images.

Population

The focus of this study is on the diagnosis and imaging of craniosynostosis within the pediatric population. Initial inclusion criteria included all patients populated by RadSearch with the search term "craniosynostosis." After this initial filter of reports, only cases with radiologist-reported evidence of craniosynostosis were included. This resulted in a study population of patients imaged between 2002 and 2022, the chronological limit of the search results when executed. Both syndromic and non-syndromic subtypes were included for analysis. Patients of all ages were included in the study cohort; however, some outliers were excluded from specific statistical analyses as outlined below.

Primary and Secondary Measures

The primary measure of this study was the subtype of craniosynostosis, including sagittal, metopic, unicoronal/bicoronal (images exported separately but statistically analyzed together), lambdoid, and complex (multi-sutural craniosynostosis). Less common subtypes observed were included in a separate category of "others," and those that did not have a subtype specified in definitive terms within the report were designated to the "unspecified" category. Secondary measures were age at radiographic diagnosis and sex as displayed in the medical record. A high-quality, representative case from each subtype was exported and their images were included in the report for additional educational benefit.

Follow-Up

Once a case fitting the inclusion criteria was identified, all instances of head and neck imaging found within RadSearch (including XR, CR, CT, and MRI) were recorded. The reason for repeat imaging was also recorded, such as post-op follow-up, surveillance, imaging for other medical conditions, etc.

Statistical Analysis

Once all cases were separated by subtype, the number of cases in each group was totaled. Additionally, the total number of male and female patients in each group was totaled. We then calculated the percentage of all cases belonging to a certain subtype, and the percentage of total cases and each subtype that were male or female. To calculate the average age at radiographic diagnosis, patients who had their first imaging performed at an age equal to or greater than 18 years were excluded, as the condition of interest was more likely an incidental finding in these patients.

Results

Patients

1811 individual exams were populated when entering the search term "craniosynostosis" into RadSearch. After a review of each associated imaging report, patients were excluded if the radiologist ruled out the condition of interest, or if evidence of the condition was equivocal. After exclusion, 628 total patients fit the inclusion criteria for this study.

Follow-up

Most of the patients observed had an initial CT scan to diagnose craniosynostosis and one or more follow-up scans for postoperative surveillance. A smaller portion had only one CT scan for initial diagnosis and no follow-up, or a series of ten or more CT scans for surveillance of more aggressive syndromic subtypes. In any case, all imaging modalities (including XR, CR, CT, and MRI) and dates/instances of follow-up imaging were collected as long as the condition of interest was visualized on imaging and reported by the radiologist.

End Points

The primary endpoint of this study was the number of patients that exhibited each subtype of craniosynostosis, including sagittal, metopic, coronal (unicoronal and bicoronal), lambdoid, and complex (multiple fused sutures). Less commonly observed subtypes were labeled as "other," including orbital, occipital, and squamosal craniosynostosis. A larger-than-anticipated proportion of patients met the inclusion criteria for the study but did not have a specific subtype of craniosynostosis listed in the report, as such, these cases were labeled as "unspecified." The results for the primary endpoint were as follows: Sagittal 33.1%, Metopic 22.5%, Coronal (unicoronal and bicoronal) 13.7%, Lambdoid 3.3%, Complex 7.3%, Other 1%, and Unspecified 19.1% (see Figure 1).

The secondary endpoints were sex as listed in the medical record and age at radiographic diagnosis. Out of the 628 patients meeting inclusion criteria, 64% of cases were male and 36% were female. Analyzing each subtype, male cases outnumbered female cases in all categories except the coronal subtype. The average age at radiographic diagnosis for all subtypes was 1 year 11.4 months. Six patients were excluded from this endpoint, as they were initially imaged at an age of or over 18 years, and the diagnosis of craniosynostosis was likely incidental in these cases.

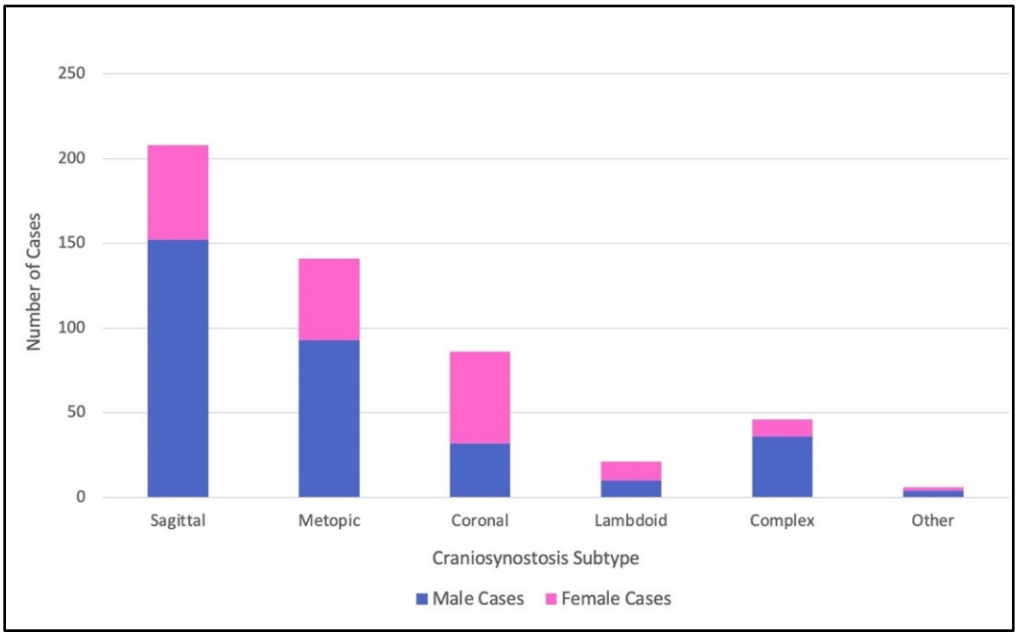

Figure 1. Craniosynostosis and their Subtypes

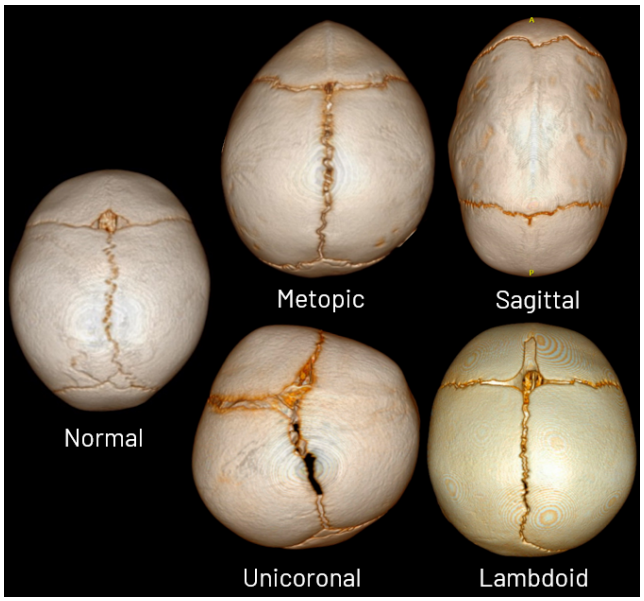

Figure 2. Superior-to-Inferior View of Different Craniosynostosis Subtypes

After statistical analysis, representative images from each subtype were exported for educational review. A broad overview of all subtypes can be seen in Figure 2, utilizing CT with 3D reconstruction to recreate the skull and its associated sutures, each displaying varying levels of patency. While abnormal skull morphology is not necessary to diagnose craniosynostosis, it strengthens the argument that early closure of a calvarial suture is indeed craniosynostosis as opposed to a mimic or other differential diagnosis. The above overview and below figures represent "textbook" examples of each subtype; notably, the patients observed in this study demonstrated a spectrum of presentations in skull morphology, with the degree of suture fusion being less variable. Representative cases of each subtype can be seen in Figures 3-7.

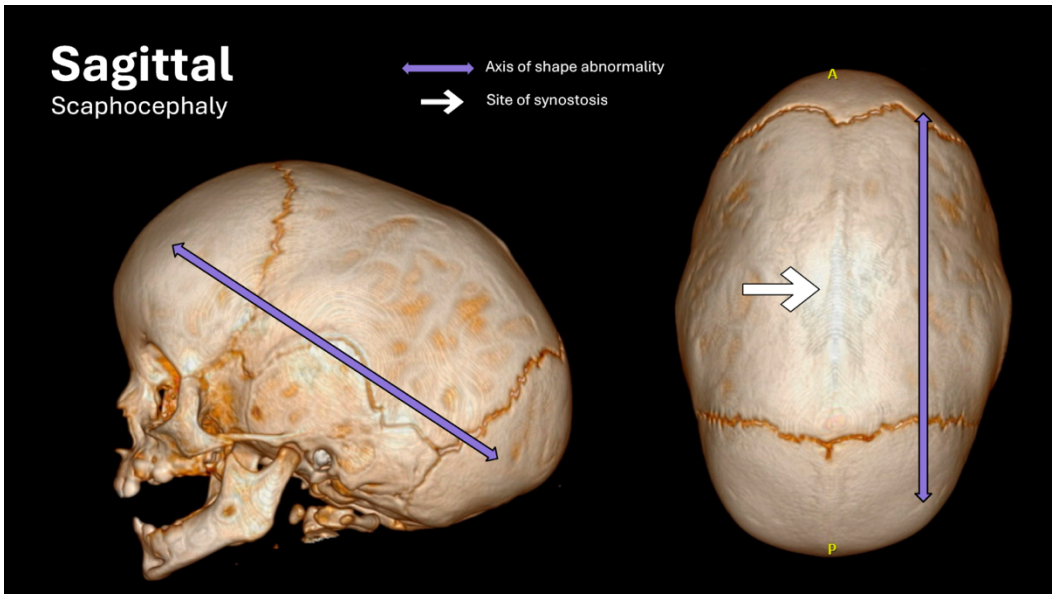

Figure 3. Representative Case of Sagittal Craniosynostosis with Associated Scaphocephaly

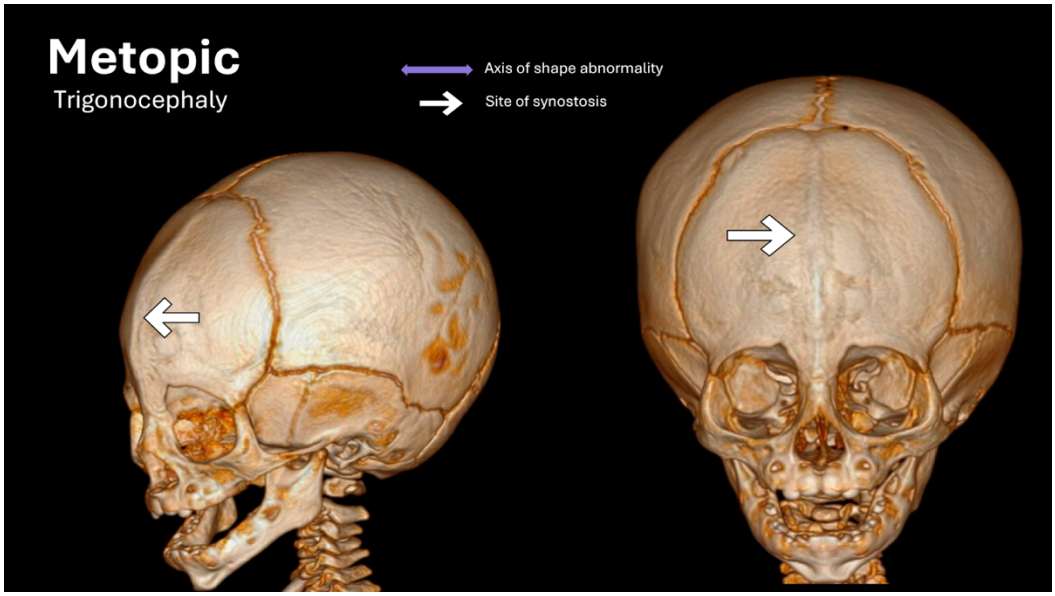

Figure 4. Representative Case of Metopic Craniosynostosis with Associated Trigonocephaly

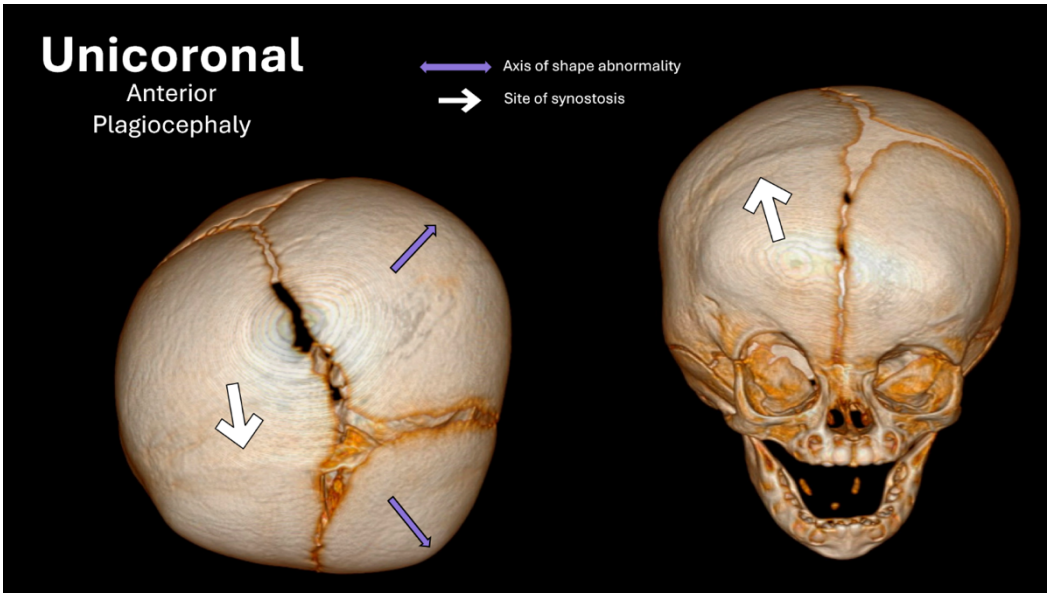

Figure 5. Representative Case of Unicoronal Craniosynostosis with Associated Anterior Plagiocephaly

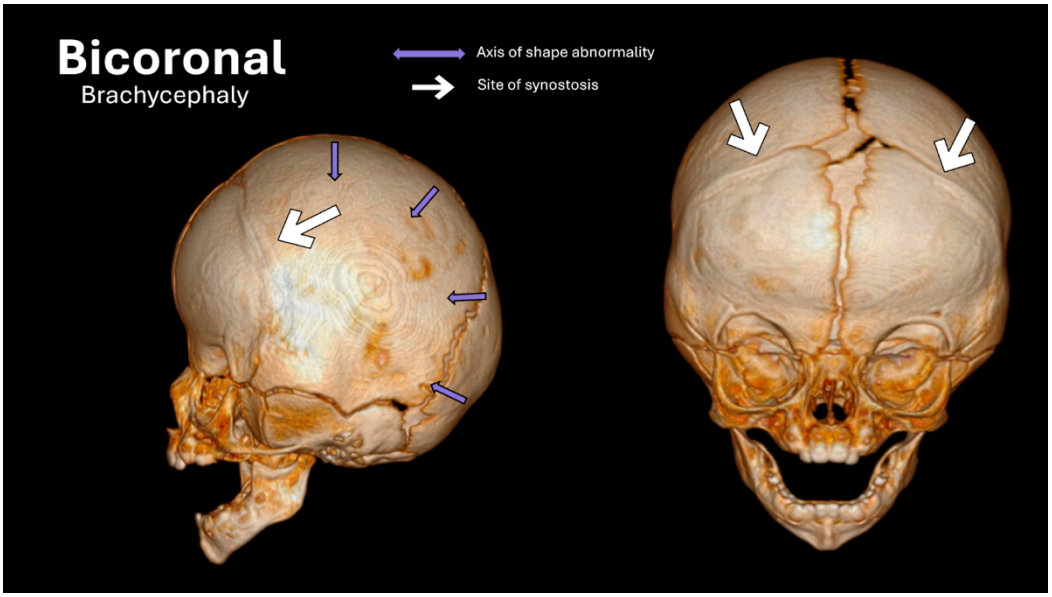

Figure 6. Representative Case of Bicoronal Craniosynostosis with Associated Brachycephaly

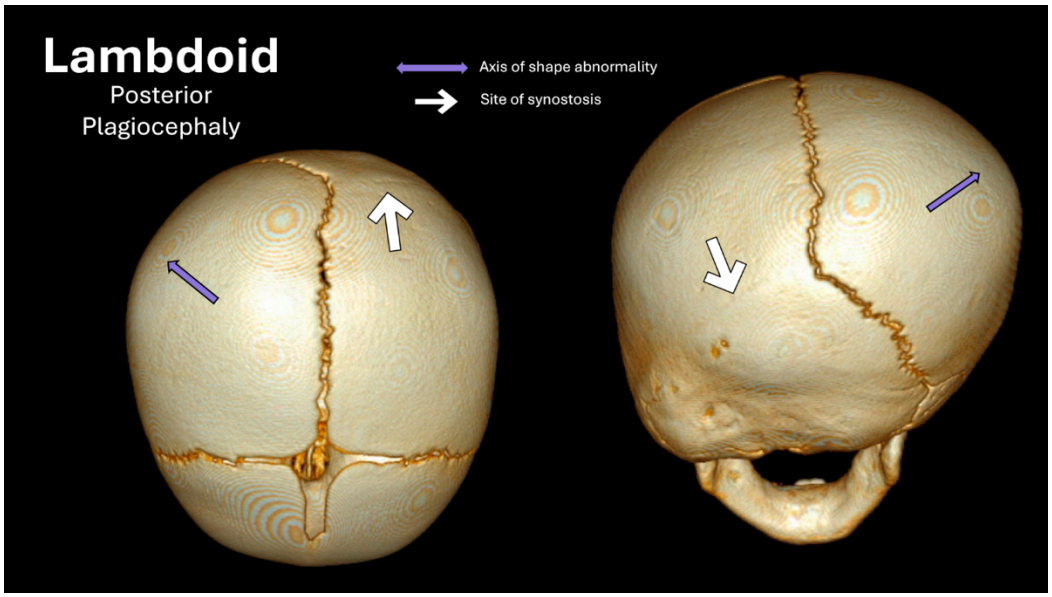

Figure 7. Representative Case of Lambdoid Craniosynostosis with Associated Posterior Plagiocephaly

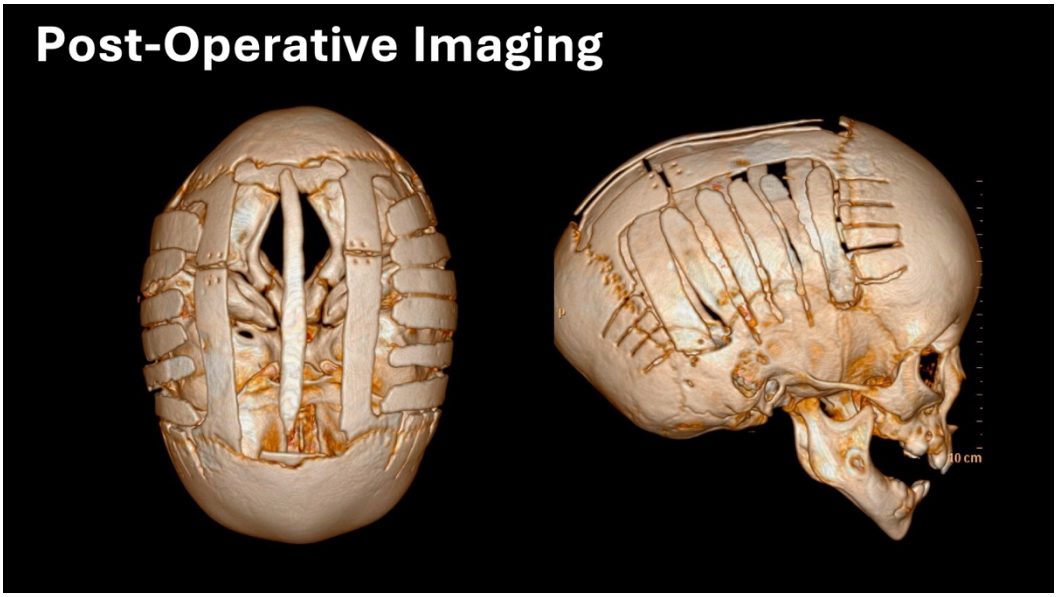

Figure 8. Post-Operative Imaging after Barrel-Stave Osteotomy for the Treatment of Sagittal Craniosynostosis

Most observed patients with uncomplicated, non-syndromic craniosynostosis chose to undergo elective surgical correction shortly after diagnosis. In this case, either endoscopic treatment for those under the age of 6 months, or open treatment with barrel-stave osteotomy (seen in Figure 8) for those over 6 months of age was performed[1]. During the follow-up period with the surgical team, one or more additional CT scans were acquired to better characterize individual postoperative recovery. In contrast, some younger patients with less severe presentations opted for non-operative treatment with helmet use only and may not have had follow-up imaging if skull morphology appeared to improve clinically.

Discussion

The results of this study closely resemble existing literature on craniosynostosis. Overall, the subtypes of craniosynostosis at our institution and their contribution to all cases match nationally reported data[1]. However, the metopic and coronal subtypes were observed to be more and less common in our population, respectively.

Age at Radiographic Diagnosis

The average age at radiographic diagnosis of 1 year 11.4 months is significantly higher than the age of clinical diagnosis within other reported studies, with many reporting less than 6 months of age[7]. 60.8% of the cohort had their first instance of imaging at less than 1 year old, and the remaining patients had their diagnostic CT between the ages of 1 to 18 years old. The scope of this project did not allow for comprehensive chart review and was limited to the content of radiology reports. More chart data would likely allow us to better understand the indication for imaging and clinical suspicion for craniosynostosis, and therefore calculate an age at clinical diagnosis, rather than solely a radiographic diagnosis. Similarly, it is likely that some of these patients had imaging acquired before the earliest indexed RadSearch results

in 2002, or transferred from outside medical systems and were initially diagnosed there.

Sex Differences

The preponderance of male-to-female cases of our cohort in a 64%/36% proportion closely mirrors existing literature, which widely supports a 2:1 male/female incidence ratio and males accounting for most midline synostosis cases. Interestingly, the only subtype that was observed to have more female cases is unilateral coronal synostosis, which was observed in an epidemiologic study in Norway[7]. The cause of this sex discrepancy has yet to be determined, although it has become an area of increased research interest within the last few years.

Treatment

The treatment of choice for most of the observed patients was elective surgical correction using either an endoscopic approach or an open approach using barrel-stave osteotomies. Endoscopic repair is the treatment of choice for infants less than 6 months old, as the skull bones are sufficiently flexible to allow endoscopic manipulation for suturectomy. In some cases, post-operative helmet use is required to allow for the proper re-ossification of the young, rapidly developing calvarium. This treatment approach typically minimizes perioperative blood loss and other complications and can result in similar aesthetic morphology, but has technical limits to the amount of reconstruction that can occur[1,8]. On the other hand, open correction is employed after the age of 6 months or with more complex and severe presentations. Barrel-stave osteotomy is commonly employed in these procedures (Figure 8) and can also be paired with post-operative helmet use to guide skull re-ossification. The goal for either surgical approach is to avoid neurological deficit as a result of increased ICP and decreased skull volume.[9] This study captured all instances of post-operative imaging as patients were being followed throughout their recovery. For non-syndromic craniosynostosis with mild presentations, sometimes a remodeling helmet was alone used to see if skull morphology would normalize non-operatively. This conservative treatment approach was also used in cases of diagnostic ambiguity, in which positional plagiocephaly had not been ruled out yet.[1]

Differential Diagnoses

When collecting data on RadSearch, the two most prevalent differential diagnoses when a clinician attempted to rule out craniosynostosis were positional plagiocephaly and metopic ridge. Positional plagiocephaly presents as an asymmetric, misshapen skull that appears like the plagiocephaly seen in either lambdoid or coronal synostosis, except that the driving factor of deformation is an external force instead of premature fusion of sutures. Labor and birth complications, comorbid conditions such as congenital muscular torticollis, and abnormal positioning of the head all can predispose an infant to non-synostotic, positional plagiocephaly.[10] While this condition is often differentiated from craniosynostosis clinically and without the need for imaging, metopic ridge is a condition that mimics the trigonocephalic skull shape seen in metopic craniosynostosis that often needs CT with 3D reconstruction to accurately characterize. Patients with metopic ridge typically present later than those with synostosis, do not meet all craniosynostosis diagnostic criteria, and have characteristic “ridging” of the metopic suture on CT as opposed to premature fusion.[11]

Limitations

With the scope of this study operating as a personal scholarly pursuit for the OHSU School of Medicine, and therefore being a lack of adequate funding and significant collaboration with other research colleagues, there are several limitations to this study that restrict its generalizability. First, only cases that had the term "craniosynostosis" listed within the imaging report were included in the study. While this is the most well-known name for the condition of interest, it is possible that some cases were missed if non-explicit report terms were used, such as "fusion of sagittal suture" or "scaphocephaly" alone. Additionally, 19.1% of cases did not have a specific subtype of craniosynostosis listed in the imaging report. Given that the author is not a trained radiologist, it was not possible to retrospectively re-classify these individuals and better understand them in the context of the primary study outcome. Another limitation was the vague diagnostic word choice in 4% of cases, which were categorized as "neutral" craniosynostosis cases as opposed to positive. These reports had descriptions such as "features of/appears similar to craniosynostosis" within the findings section, but without clear language in the interpretation to confidently say that they meet diagnostic criteria for craniosynostosis, and therefore inclusion criteria for the study. Finally, the most significant limitation of this study is the single-institutional patient recruitment and the lack of data collection of variables regarding morbidity and mortality. These factors decrease this study's overall generalizability to the pediatric population.

Future Direction

As the scope of this study did not include morbidity or mortality data of the studied craniosynostosis patients, and generally did not recruit from enough clinical sites to create meaningful statistical power, this would be a logical future direction for this project. Work could be performed to fill these local data gaps and take steps to update or compare to existing prevalence and incidence metrics for this condition.

Conclusions

This retrospective, single-institutional study aimed to better characterize a local population of craniosynostosis cases from a radiographic perspective. 628 patients were identified as having evidence of this condition based on imaging reports indexed by RadSearch, which included a complete fusion of cranial sutures and resultant calvarial deformities. The subtypes of craniosynostosis at our institution and their contribution to all cases, as well as the male-to-female incidence ratio of 2:1, match nationally reported data. However, the metopic and coronal subtypes were observed to be more and less common in our population, respectively. The gold standard for diagnosing craniosynostosis in this study was CT with 3D reconstruction, which was also utilized to provide follow-up imaging after post-operative cranial vault reconstruction or non-operative helmet use. Positional plagiocephaly and metopic ridge are conditions that appear morphologically similar to craniosynostosis but can be ruled out by careful history taking, physical exam, and CT scans if needed. There are still numerous gaps in knowledge regarding this condition, including its true non-syndromic pathogenesis and preponderance to affect males over females.

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