Uvular Malformation in the Presence of Deformational Plagiocephaly

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Background: Deformational plagiocephaly is cranial asymmetry caused by external forces on the skull. Deformational plagiocephaly is seen in 5% to 48% of healthy newborns. Incomplete uvular fusion, in contrast, is one of many uvular malformations. The incidence of all degrees of incomplete uvular fusion is approximately 1% in healthy children. Bifid uvula is a malformation that is often considered a microform cleft palate or a marker for submucous cleft palate.

Methods: This is a retrospective study of patients with deformational plagiocephaly seen at the Upstate Cleft and Craniofacial Center between January 1, 2006, and September 30, 2011. Patients were identified by the *International Classification of Diseases, Ninth Revision* code for plagiocephaly. Seventy-nine patients were excluded with craniosynostosis and syndromic diagnoses. One hundred forty-six patients with deformational plagiocephaly were included in the study. Data were collected for sex, age at presentation, parity, multiple births, delivery, oligohydramnios, cephalohematoma, uterine abnormalities, fetal position, and intrauterine growth restriction. Clinical findings were collected including location of cranial flattening and uvular malformations.

Results: Twenty-four of 146 patients with deformational plagiocephaly had incomplete fusion of the uvula ranging from complete bifid uvula to a notched uvular tip (16.4%). This association was statistically significant (odds ratio, 18; 95% confidence interval, 11.1–28.9). Most patients (62.3%) were male. We recorded primiparity (44.5%), multiple births (17.1%), vacuum-assisted delivery (6.2%), cesarean section (36.3%), oligohydramnios (4.1%), uterine abnormalities (2.1%), abnormal fetal position (3.4%), and intrauterine growth restriction (1.4%). Ten of the 24 patients with plagio-cephaly and uvular malformation were seen for an initial consultation only in our chart system. Of the remaining 14 patients with follow-up, none had recorded signs or symptoms of velopharyngeal insufficiency.

Conclusions: The incidence of incomplete uvular fusion in infants with deformational plagiocephaly is 16.4%, which is significantly higher than the approximate 1% incidence reported in the general population. This is the first report of uvular malformation in the presence of deformational plagiocephaly.

Key Words: Uvular malformation, deformational plagiocephaly

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Deformational Plagiocephaly

Plagiocephaly is a general term used to describe cranial asymmetry. It is derived from the Greek words *plagios*, meaning "oblique", and *kephalê*, meaning "head".[1] Deformational plagiocephaly is caused by intrauterine and/or postnatal external forces that deform the skull. In contrast, synostotic plagiocephaly is caused by premature fusion of the cranial sutures.[2] Deformational plagiocephaly is more common than synostotic plagiocephaly, with a prevalence of 5% to 48% of healthy newborns.[1] The diagnosis of posterior deformational plagiocephaly can often be made by physical examination alone. It is characterized by varying degrees of abnormal cranial features including asymmetrical parallelogram skull shape, unilateral parieto-occipital flattening, ipsilateral frontal bossing, and anterior displacement of the ipsilateral ear (Fig. 1).[3]

Risk Factors

Multiple risk factors for deformational plagiocephaly have previously been identified. Peitsch et al[4] identified assisted vaginal delivery, prolonged labor, birth position, primiparity, and male sex as risk factors. Kane et al[5] reported that deformational plagiocephaly is associated with fetal constraint: small uterus, large fetus, oligohydramnios, and multiple-birth pregnancies.

After reports that infants who slept in the prone position were at an increased risk of sudden infant death syndrome, the American Academy of Pediatrics (AAP) Task Force on Infant Positioning and Sudden Infant Death Syndrome formally recommended that healthy infants be placed supine or on their sides.[6] The Back to Sleep Campaign has been one of the most successful public health initiatives. After this campaign, the incidence of prone sleeping decreased from 70% in 1992 to 17% in 1998, and the incidence of sudden infant death syndrome decreased from a rate of 1.2 cases per 1000 live births in 1992 to 0.72 cases per 1000 live births in 1998.[7,8] A tertiary care craniofacial center reported a 5- to 6-fold increase in referrals for deformational plagiocephaly between 1992 and 1994 compared to the previous 13 years, suggesting an association with the new AAP recommendations for infant positioning.[5]

Uvular Malformations

Bifid uvula is a uvular malformation that is often considered a microform cleft palate or a marker for submucous cleft palate (SMCP). A complete bifid uvula is less common than incomplete (partial) fusion of the uvula.[9] Other uvular malformations include a broad uvula and an abortive cleft uvula. The incidence of incomplete uvular fusion or bifid uvula is approximately 1%.[9,10] Meskin et al[9] studied incomplete uvular fusion in 2 general populations and

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FIGURE 1. Infant with posterior deformational plagiocephaly.

classified the malformations based on the percent length of the incomplete fusion. He noted a 1.34% and 1.47% incidence of all degrees of bifidity in these 2 populations. Of note, most of these patients (88% and 82%, respectively) did not have a complete bifid uvula but rather had less than 25% length of bifidity. Shprintzen et al[11] reported an incidence of uvular abnormalities in a busy suburban pediatric practice of 2% to 4%. Uvular abnormality was described as either notched, bifid, or broad. However, the incidence of each uvular abnormality was not given.

The diagnostic criteria for SMCP was first defined by Calnan[12] as a clinical triad of bifid uvula, zona pellucida, and a notch in the posterior border of the hard palate (Fig. 2). The incidence of SMCP is between 0.02% and 0.08%.[10,13] Kaplan[14] introduced the term "occult SMCP" to describe patients who had velar abnormalities but did not exhibit any of the anatomical signs of the classic triad. The diagnosis of an occult SMCP can only be made by nasopharyngolaryngoscopy to document a lack of velar eminence on the nasal surface of the soft palate, causing it to appear flat or concave. This appearance suggests absence of the uvula muscle and diastasis of the velar muscles.

Velopharyngeal insufficiency (VPI) occurs in approximately 10% of patients with SMCP because the hypoplastic and aberrantly inserted levator palatini muscles do not permit the soft palate to elevate sufficiently during speech, leading to incomplete velopharyngeal closure.[10] Submucous cleft palate is often diagnosed late when a patient develops abnormal speech owing to velopharyngeal insufficiency.[15] This is characterized by hypernasal resonance and decreased intraoral pressure during speech.[15] In young patients who have not yet developed speech, velopharyngeal insufficiency can present as feeding difficulties, such as nasal regurgitation.

Malformation in the Presence of Deformational Plagiocephaly

At the Upstate Cleft and Craniofacial Center, every child undergoes an oral examination with documentation of uvular malformations, if present. We have frequently observed uvular malformations in patients with deformational plagiocephaly. These malformations include all degrees of incomplete uvular fusion from grooved or notched uvular tips to complete bifidity. Our study's aim was to determine if there is a higher incidence of incomplete uvular fusion in patients with deformational plagiocephaly compared to the general population. To date, there have been no reports of uvular malformations in the presence of deformational plagiocephaly.

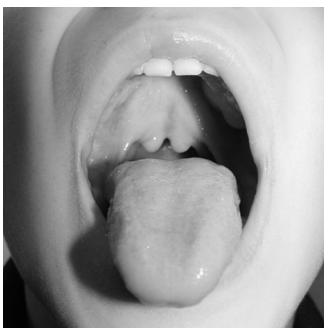

FIGURE 2. Submucous cleft palate. Patient has zona pellucida and bifid uvula present on physical examination.

PATIENTS AND METHODS

Institutional review board approval was obtained. We performed a retrospective chart review of 225 consecutive patients seen at the Upstate Cleft and Craniofacial Center between January 1, 2006, and September 30, 2011, with plagiocephaly. Patients were identified using the *International Classification of Diseases, Ninth Revision* code for plagiocephaly. Exclusion criteria included patients with craniosynostosis and those who did not meet diagnostic criteria for deformational plagiocephaly. Posterior deformational plagiocephaly was diagnosed by the presence of unilateral parieto-occipital flattening often with ipsilateral frontal bossing, asymmetric parallelogram shape, and anterior displacement of the ipsilateral ear. Wide variation in severity classification in the charts precluded our ability to consistently gather these data.

After exclusion, we identified 146 patients with deformational plagiocephaly. The mean age at presentation and diagnosis was 8.2 months, with a range of 1 week to 83 months. Data were collected for sex, parity, maternal uterine abnormalities, fetal position, intrauterine growth restriction (IUGR), cephalohematoma, oligohydramnios, assisted delivery, cesarean section, and singleton versus multiple-birth pregnancies. All patients had an oral examination. Physical features were collected including location of cranial flattening (right vs left; anterior vs posterior), uvular malformation, presence of torticollis, and other abnormalities. A uvula was considered to have incomplete fusion if there was any degree of clefting or groove present in the tip. Other uvular malformations including broad shapes and short uvulas were not included in this analysis. Control group data were derived from the prevalence of bifid uvula in the large random population study by Weatherly-White in 1972, which is consistent with population studies by Meskin including all degrees of incomplete uvular fusion.[9,10]

RESULTS

Of 146 patients with deformational plagiocephaly, 24 patients (16.4%) had a degree of incomplete uvular fusion. This association was highly statistically significant (odds ratio, 18; 95% confidence interval, 11.1–28.9; risk ratio, 16; 95% confidence interval, 10.2–23.7). Sixty-two percent of patients (91/146) were male (Table 1). The wide range of age at presentation highlights the many causative factors of deformational plagiocephaly including in intrauterine fetal constraint (congenital deformational plagiocephaly) and postnatal deformation (acquired deformational plagiocephaly).

TABLE 1. Demographics

Total No. of Patients	**146**
Mean age at first office visit, mo	8.2
Age range at first office visit, mo	0.2–83
Sex	
Female	55
Male	91

mo, month.

TABLE 2. Previously Described Risk Factors for Deformational Plagiocephaly

Risk Factors	No. of Patients	Percentage (%)
Primiparity	65	44.5
Vacuum-assisted delivery	9	6.2
Cesarean section	53	36.3
Oligohydramnios	6	4.1
Multiple births	25	17.1
Uterine abnormalities	3	2.1
Abnormal fetal position	5	3.4
Intrauterine growth restriction	2	1.4

All of the patients had posterior deformational plagiocephaly. Forty-eight patients (35%) had left deformational plagiocephaly, and 88 patients (65%) had right deformational plagiocephaly. Sidedness data were not available for 10 patients. Patients with deformational plagiocephaly were most commonly born to primipara mothers (44.5%). We identified multiple births (17.1%), vacuum-assisted delivery (6.2%), cesarean section (36.3%), oligohydramnios (4.1%), uterine abnormalities (2.1%), abnormal fetal position (3.4%), and intrauterine growth restriction (1.4%) (Table 2). Co-occurring deformational anomalies were identified including clubfeet (0.7%), hip dysplasia (0.7%), and torticollis (46.6%) (Table 3). Ten (42%) of the 24 patients with incomplete uvular fusion were seen only once in our chart system. Of all 24 patients with incomplete fusion, none had recorded signs or symptoms of velopharyngeal insufficiency.

DISCUSSION

Co-occurring Malformation and Deformation

There are reports of congenital deformations co-occurring with deformational plagiocephaly including congenital hip dislocation, bat ears, congenital scoliosis, clubfeet, hip dysplasia, auricular anomalies, and mandibular hypoplasia.[4,15,16] However, this is the first report of uvular malformation in patients with deformational plagiocephaly. We found a significantly higher incidence of incomplete uvular fusion in patients with deformational plagiocephaly compared to the reported incidence in the general population, 16.4% and approximately 1%, respectively. We also found a high percentage of previously identified risk factors for deformational plagiocephaly in our patients including male sex, multiple-birth pregnancy, and primiparity. Severity data of deformational plagiocephaly was not available for our chart review.

Data from a large population study of healthy children by Weatherly-White was used as a control population for analysis.[10] The results of this study by Weatherly-White are consistent with 2 other population studies by Meskin et al[9] who reported the incidence of all degrees of incomplete uvular fusion as 1.34% and 1.47%. Because there is no control group for this study's patient population, there may be a larger than 1% incidence of incomplete uvular fusion in the nonplagiocephaly population using the criteria in this study. Furthermore, the incidence of incomplete uvular fusion in our patient population may be greater than the incidence in mild cases of plagiocephaly not referred to a craniofacial center.

TABLE 3. Other Anomalies Present With Deformational Plagiocephaly

Other Anomalies	No. of Patients	Percentage (%)
Incomplete uvular fusion	24	16.4
Torticollis	68	46.6
Cephalohematoma	1	0.7
Clubfoot	1	0.7
Hip dysplasia	1	0.7

Risk Factors

On average, male infants have a larger head circumference than girls.[17] A larger head circumference may be at higher risk of deformation, especially in the setting of uterine crowding such as multiple births, uterine fibroids, or oligohydramnios. Furthermore, males are less flexible than females, which makes them more susceptible to deformation.[18] Deformational plagiocephaly, likewise, is more common in males than in females. The literature reports male-to-female ratios between 2:1 and 3:2.[4,16] Our study revealed a 3:2 male-to-female ratio.

Seventeen percent (25/146) of our patients with deformational plagiocephaly were the result of a multiple-birth pregnancy rather than a singleton birth. Previous literature has reported that the prevalence of plagiocephaly in twins was 55.6%, more than 4 times as frequent as in singletons.[4]

Previous studies have observed a 4% incidence of oligohydramnios in infants with deformational plagiocephaly, but the numbers were too small to draw any conclusions.[4] We also found a 4.1% incidence of oligohydramnios in infants with deformational plagiocephaly. Oligohydramnios may lead to decreased fetal movement and/or fetal compression, which would seemingly increase the risk of cranial deformation.

Deformational posterior plagiocephaly has been shown to be more common on the right than the left.[4,16] In our study, right-sided deformational plagiocephaly was present in 65% of the patients. It has been reported that infants turn their heads to the right side nearly 80% of the time.[19] In addition, left occipital anterior presentation of infants in the birth canal is common.[4] Descent through the birth canal in this position involves right occiput compression against the maternal pelvic bone.[4] Our clinical records in this retrospective chart review did not consistently include presentation of the baby at birth.

Given that almost all newborns today sleep supine, one may expect most newborns to develop posterior deformational plagiocephaly. However, in actuality, only a minority of newborns have this condition.[1] This group might have a factor causing susceptibility to deforming forces. Michels et al[20] reported that women who took greater than recommended doses of folic acid during pregnancy gave birth to a higher incidence of children with deformational plagiocephaly. In 2012, Michels et al[21] reported that twice the recommended daily intake of folic acid supplementation occurred significantly more often among the mothers of children with deformational plagiocephaly compared with mothers of children without deformational plagiocephaly. This association suggests a possible detrimental effect of elevated prenatal folic acid levels on skull development. However, no biological pathway linking plagiocephaly and folic acid has been found to prove causality. This previously reported correlation does not change the importance of folic acid supplementation to prevent neural tube defects.

In the literature, maternal folate is associated with a decreased incidence of cleft lip with or without palate.[22] The literature is not conclusive, however, as studies also have shown no protective effect of maternal folate on cleft lip with or without cleft palate or cleft palate alone.[22] Previous data have shown an association between increased maternal folate intake and an increased incidence of bifid uvula.[20,21] If maternal folate has a protective effect on clefting, we hypothesize that children with incomplete uvular fusion may have been genetically or environmentally predisposed to complete palatal clefting. In these cases, folate may have had a protective effect, resulting in only a microform cleft palate or uvular malformation rather than a complete palatal cleft.

Given the high percentage of our patients with uvular malformation and deformational plagiocephaly, it is possible that there are shared conditions causing increased susceptibility to both, including a genetic disruption, environmental insult, or a combination of both. These postulations reference previous studies, and no studies have been done specifically showing any biological link between uvular malformation and deformational plagiocephaly.

Velopharyngeal Insufficiency

Bifid uvula is considered a marker for SMCP. Shprintzen et al[11] found that of 25 children with bifid uvula, all but 2 children had some or all of the landmarks of SMCP on nasopharyngolaryngoscopy. Approximately 10% of children with SMCP have speech abnormalities owing to velopharyngeal insufficiency (VPI).[10] Although speech abnormalities are usually mild and respond to speech therapy, up to 10% of VPI can be severe and may require surgical intervention.[10] This report highlights the importance of a thorough oral examination to look for uvular malformations in infants with deformational plagiocephaly as this may be a marker for SMCP. Early recognition of uvular malformations will allow for parental counseling about the risk of VPI and abnormal speech development associated with SMCP.

At the Upstate Cleft and Craniofacial Clinic, surgical management for velopharyngeal insufficiency is not considered until there is sufficient speech to evaluate. At that point, a Furlow palatoplasty is considered at 4 years of age. If the patient is older than 4 years, a pharyngeal flap may be performed, depending on the size of the gap.

Of patients with isolated bifid uvula, the risk of VPI is low and surgical treatment is not necessary. In our study, 10 (42%) of the 24 patients with incomplete uvular fusion were seen for an initial consultation only. Of all 24 patients, none had recorded signs or symptoms of velopharyngeal insufficiency. The absence of velopharyngeal insufficiency in this population renders it impossible to interpret further with the small study power.

In conclusion, this is the first report of uvular malformation in the presence of deformational plagiocephaly. We observed a significant association between incomplete uvular fusion and deformational plagiocephaly. Future research is ongoing to examine the relationship among deformational plagiocephaly, uvular malformations, and maternal folic acid intake.

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