Nasofrontal encephalocele in a child, associated with a nasal dermal sinus in his mother: familial cranial malformations. Illustrative case

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BACKGROUND Dermal sinuses and encephaloceles are uncommon central nervous system malformations. The occurrence of these malformations in different members of a family is very rare.

OBSERVATIONS This report documents the unique case of a 10-month-old boy with a frontonasal encephalocele, born to a mother with a nasal dermal sinus. These two specific neural tube defects have not been previously reported as occurring between parent and child.

LESSONS This case demonstrates potential heritable links in central nervous system malformations through the occurrence of a frontonasal encephalocele in a child born to a mother with a dermal sinus. The connection between these two malformations in a heritable manner suggests a potential need for further research into the genetic pathogenesis of such defects to predict them more accurately within families.

Illustrative Case

History and Presentation

A 10-month-old boy was admitted with a bulging mass in his frontonasal area. The lesion was evident at birth without any prenatal diagnosis. It decreased in size when the child was at rest and in a supine position but increased in size during crying and straining. He is the second child of non-consanguineous parents with no significant medical history. His prenatal care was delivered according to standard care practices.

During the physical examination, the child displayed normal neurodevelopmental milestones. The mass was soft with intact skin and no tenderness (Fig. 1). Brain magnetic resonance imaging (MRI) was performed, revealing a frontonasal encephalocele with communication between intracranial and extracranial spaces through a bone defect in the frontonasal area (Fig. 2).

Operation and Outcomes

The child underwent bifrontal craniotomy in a supine position, during which we dissected the neck of the encephalocele sac, cut the

ABBREVIATIONS MRI = magnetic resonance imaging; MTHFR = methylenetetrahydrofolate reductase; NTD = neural tube defect.

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communication with the extracranial part of the encephalocele, and repaired the dura. There was a 1 × 1–cm bone defect in the midline part of the frontal bone in front of the crista galli, and the dural sac was herniated through this defect to the frontonasal area. The bone defect was covered with a same-size bone flap harvested from the posterior part of the frontal bone. The soft tissue mass in the nasion was removed with an elliptical incision around the neck of the sac, and the incision was repaired with 5.0 sutures. The postoperative period was uneventful.

The patient’s mother had had a nasal dermal sinus since birth, with a small black hair on the orifice of the sinus and no discharge (Fig. 3). There was no previous infection or bulging around the sinus; therefore, she did not undergo further investigation.

**Patient Informed Consent**

The necessary patient informed consent was obtained in this study.

**Discussion**

The cranial dysraphisms include different severities of malformations comprising cranial dermal sinuses, meningoceles, and encephaloceles. Encephaloceles and cranial meningoceles are caused by mesodermal aberrations and associated cranial bone defects that provide cerebral contents or only meningeal herniation through these defects, respectively. Encephaloceles are considered as NTDs by some authors or as post-neurulation abnormalities. The association of encephaloceles with other congenital nervous system malformations in the same persons or in their families has been described before. Dermal sinuses can be found at any part of the neuraxis, though rarely in the nasal area, and can result from an imperfect separation of the ectoderm and neuroectoderm during the 3rd to 5th weeks of gestational age. The association of a dermoid cyst and an encephalocele in a patient has been reported by Tokmak et al. They described a 15-year-old girl with a painful mass at the base of her mouth in the upper neck, which was found to be a dermoid cyst. Craniocervical MRI for more investigation of that lesion confirmed an occipital bony defect with a cerebellar encephalomeningocele. The occurrence of a dermal sinus with another congenital abnormality, including a dermal sinus, myelomeningocele, and meningocele in two persons of a

**FIG. 1.** Photograph of the child with an anterior encephalocele, presenting with a soft tissue mass in the upper portion of the nose.

**FIG. 2.** Brain MRI confirmed the relation between the intracranial space and the nasal area.

**FIG. 3.** Photograph of the mother shows a nasal dermal sinus (right) together with a postoperative image of her son (left).
family, has been reported before. Ansari et al. reported a 2-year-old boy with a complicated occipital dermal sinus and intracranial dermoid tumor and abscesses. His mother had an asymptomatic occipital dermal sinus. These authors proposed a genetic basis for certain instances of occipital dermal sinuses. However, there is no report of the association of an anterior encephalocele and a dermal sinus in two members of one family.

The cause of the encephalocele and dermal sinus in first-degree members of a family is unknown, but some explanations can be considered. Prior studies have suggested that a multifactorial inheritance model may be the culprit of such defects, with a specific focus on the MTHFR folate metabolism gene. Although environmental factors may play a key role in the development of NTDs, such as a lack of access to folic acid or prenatal care, this gene abnormality shows that NTDs can occur even if an individual has access to such resources. Therefore, possible genetic mechanisms of NTDs must be explored as the reasons for an increased incidence of these defects in a family.

One study has shown that a thermolabile form of the MTHFR enzyme had reduced folate metabolic activity, but this deficiency could be supplemented by folic acid. Another study has shown that the same mutation that leads to the thermolabile MTHFR enzyme (677C>T) is associated with a significant risk of developing an NTD in utero. Meta-analyses have also shown that the MTHFR 677TT genotype is a risk factor for NTDs in mothers (50%–70% increase) and their children (80%–90% increase). These studies suggest that the mysterious origins of NTD inheritance patterns can be elucidated by further investigations into polymorphisms of the MTHFR gene.

A female predominance for these malformations may confirm a genetic component. In addition, a relatively high rate of association of dermal sinus with Klippel-Feil anomaly, which has a genetic basis, can explain the genetic mechanisms causing these anomalies. Moreover, an abnormal karyotype like trisomy 18 and 13 has been associated with some kinds of NTDs and encephaly.

Observations

The different levels of severity between the mother and the child in our case show the random nature of these malformations and the importance of elucidating genetic mechanisms so that families can prepare for the potential complications of a defect in their child. Management of these conditions can vary from child to child, based on the level of severity of the malformation. Such variations in malformations have been previously documented by Kankam et al. through a scoring system and also highlight the importance of accurately diagnosing the severity of an encephalocele or any other malformation.

Lessons

This case report of a 10-month-old boy with a frontonasal encephalocele and his mother with a nasal dermal sinus represents a familial occurrence of central nervous system malformations. The association of these defects in first-degree relatives in this case strongly suggests that there is a common genetic component as previously discussed, such as the multifactorial inheritance model or an MTHFR gene mutation. Genetic counseling for individuals with a family history of NTDs can be explored as an option during family planning. Ongoing research and discovery in the field of NTDs can also shed light on the seemingly unpredictable nature of these defects. Moreover, parents can be better informed about any potential health challenges that their newborn may face.

References


Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: El Khashab, Nejat. Acquisition of data: Nejat. Analysis and interpretation of data: El Khashab, Ali. Drafting the article: El Khashab, Ali. Critically revising the article: El Khashab, Ali, Nejat. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: El Khashab. Administrative/technical/material support: El Khashab, Nejat. Study supervision: El Khashab, Nejat.

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