



REVIEW ARTICLE

Congenital Dermal Sinus: case series and the consequences of late diagnosis and treatment

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Abstract

Introduction: Congenital Dermal Sinus (CDS) is a rare closed dysraphism that can present throughout the extent of the neuroaxis. It occurs due to a failure in the embryogenic development, most commonly at the lumbar region. Clinical presentation of CDS usually consists of cutaneous stigmas that have the potential to be diagnosed at birth. However, the majority of patients are diagnosed older and after complications such as meningitis, abscess, osteomyelitis or rupture of an associated epi/dermoid cyst. Complete exeresis is the definitive treatment.

Case report: The authors experienced 3 cases of CDS. Of the 3 cases, 1 was at lumbar level and 2 were at the cranial level (occipital and nasal regions, respectively). Although all of them presented at a different location, they were all diagnosed due to a history of infection. The first case describes a patient with an enlarged occipital tissue, loss of developmental milestones and hydrocephalus. The second case describes repeated infection of the cutaneous stigma (lumbar dimple), difficulty in walking and severe intraspinal infection. The last case, more rare, depicts a patient with a hairy pit in the nasal region and recurrent periorbital cellulitis. All cases presented complications, being occipital dermoid cyst and hydrocephalus; severe intraspinal empyema and frontal dermoid cyst, respectively. All patients progressed well after aggressive surgical treatment and prolonged antibiotic therapy.

Key words: congenital dermal sinus, dermal sinus, dermal sinus tract, occult dysraphism, cutaneous stigma

Introduction

Congenital Dermal Sinus (CDS) is a rare, closed dysraphism that can present throughout the extent of the neuroaxis (1–6). It occurs during primary neurulation as a failure of the ectodermal disjunction, which is incomplete, leaving a stalk that communicates the cutaneous and neural ectoderm. The prevalence of CDS of all types has been estimated to be 1 per 2,500 live births, up to 63% in the lumbar region. More than half of the cases are associated with dermoid or epidermoid tumors (6–8).

As a benign lesion with normal neurological status at birth, the initial clinical presentation of CDS usually consists of cutaneous stigmas like dimples, which has the potential to be diagnosed early in the childhood. However, as CDS has a true lumen that communicates with a cutaneous opening and is the route of entry for pathogens, most patients are diagnosed after infected complications, such as intraspinal abscess or meningitis (5,6,9). Therefore, once diagnosed, CDS requires urgent surgical removal of the tract even in asymptomatic patients to prevent these potential infections (4,7,10).

The goal of this study is to present a series of 3 cases of CDS, to illustrate the extent of the disease and to promote awareness of the importance of early diagnosis and treatment.

Case Report

Case 1

One-and-a-half-year-old girl with a growing occipital bulging since birth, initially diagnosed as a sebaceous cyst, and normal psychomotor development until 10 months of age, when she lost the ability to stay up with support and crawl. The mother reported recurrent infections of the cutaneous stigma associated with febrile episodes, but without further investigation. At age of 1 year and 5 months, she presented at the emergency unit of another institution with fever, lethargy, and vomiting. A CT scan showed

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tetraventricular hydrocephalus, and she was submitted to external ventricular drainage. She evolved with an eruption of the occipital lesion and was referred to our pediatric unit. New CT-scan was performed and showed an occipital midline bone discontinuity and a well-defined isodense round intra-axial mass associated to hydrocephalus. MRI was compatible with the diagnosis of occipital dermal sinus with an associated ruptured dermoid cyst (Figure 1, A-C). We performed a suboccipital approach and encountered the invasion of skull bone and dura. Sinus excision was made and the dermoid cyst was well-defined, cystic and was partially resected due to brainstem extension. The bacterial culture result was negative and after operation the patient was administered empiric systemic antibiotic therapy for 4 weeks and a VP-shunt was placed. After multidisciplinary treatment, she was discharged with improvement of the ability to walk, but persisted with ataxia. No further complications were reported.

Case 2

Boy with 2 years and 10 months of age, with repeated left periorbital cellulitis, presented with a new episode and frontal swelling over a small hairy pit in the nasal region, that he had since birth (Figure 1, D). During the investigation, he was submitted to a CT-scan and a MRI, which showed a periorbital abscess, osteomyelitis of the frontal bone, patent foramen cecum, and a dermoid cyst adjacent to the dura. The patient was submitted to a bicoronal zig-zag incision, and a low based bilateral frontal craniotomy of approximately 4x8cm² was performed. This allowed direct visualization of the intracranial part of dermal tract and, with little brain retraction, it was possible to see the end of the tract on an infected dermoid cyst extending to the supero-medial aspect of the left orbit. After drainage and washing of the abscess, we performed a small durotomy that showed that the dermoid cyst was in between the layers of the dura. Then, a small osteotomy in the shape of a triangle, of approximately 3cm in each side, was done, for an “en bloc” resection. The dura was latter sutured. This allowed for a complete resection of the dermoid cyst and of the dermal sinus (Figure 1, E-F). Culture was positive for *Staphylococcus methicillin* sensible. After finishing the antibiotic therapy he was discharged with no other complaints.

Case 3

3-year-old girl, presenting a lumbosacral cutaneous stigma characterized by hypertrichosis, hemangioma and a small dimple (Figure 1, G), without previous medical history. Despite the different stigmas presented, they were never considered suspicious. After two years of age, the patient began

to experience frequent falls and never managed sphincter control. The patient was then referred to our department, being identified with an hyperactive neurogenic bladder, in addition to important distal paresis of the left lower limb. MRI showed extensive intrarachial empyema, with important arachnoiditis, involving the entire final portion of the spinal cord, including the medullary conus and the cauda equina. It was also possible to visualize a fine tract that extended from the dimple to the spine. The patient underwent surgery on an emergency basis, with extensive laminotomy from L1 to L5, and it was possible to identify the sinus tract with dural extension. After dural opening, a huge empyema was identified and drained, in addition to microscopic dissection of the arachnoiditis presented. The culture of the collected material did not show germs. Broad-spectrum antibiotic therapy was performed. Despite all efforts, the patient showed no motor or urinary improvement.

Discussion

Introduced by Earl Walker and Paul Bucy in 1934, the term congenital dermal sinus (CDS) refers to a congenital malformation forming an epithelial tube from the skin to the central nervous system and/or its coverings. It consists of a closed dysraphism caused by an incomplete disjunction of the neuroectoderm and the cutaneous ectoderm during primary neurulation, around the 4th week of fetal development (1–6,8).

Differential diagnosis must include Limited Dorsal Myeloschisis (LDM), another congenital defect of the primary neurulation. In the separation of neuro and cutaneous ectoderm, mesoderm cells interposition form a solid tract. In imaging studies, mainly MRI, it may be possible to see the absence of lumen in the tract in LDM, which is commonly present in CDS. In contrast with LDM, which usually presents with tethered cord syndrome, CDS can present with infection because of the presence of a lumen in its tract allowing contact of external pathogens with the central nervous system. For that reason, it configures a surgical emergency (9).

With prevalence estimated in 1 per 2,500 live births, CDS can occur in the midline in the whole extent of neuroaxis, and more than half are associated with a dermoid or epidermoid cyst (6–8). There is no consensus in the literature regarding its dispersion in the neuroaxis. During literature review, Powel et al (3) found 63% of CDS in the lumbar region, and 27% in the occipital, while Kalkn et al (8) found 41% in the lumbar region. Nevertheless, it all points to the lumbosacral region as the most common location, and cervical as the last one (6).

Although considered a benign lesion that presents with normal neurological status at birth, in about half

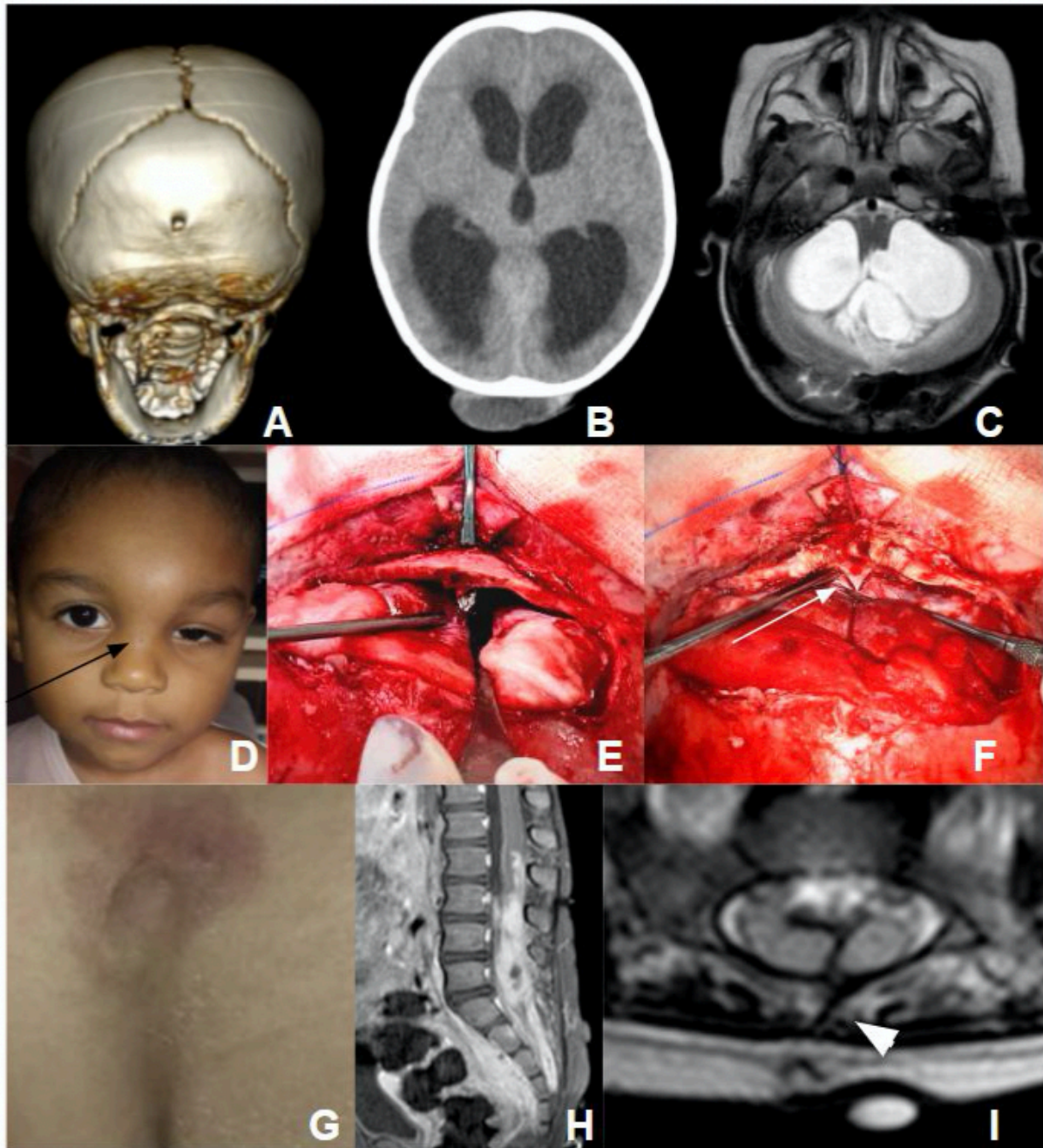


Figure 1 - Cases of CDS. Case 1 (A-C), A) 3D CT-scan reconstruction with occipital bone defect where the CDS tract was passing through, B) Axial CT-scan with hydrocephalus, C) Axial T1 MRI showing dermoid cyst in the posterior fossa. Case 2 (D-F) D) Presentation with left ptosis due to periorbital cellulitis and frontal building over a small hairy pit (black arrow) in the nasal region. E) Frontal craniotomy with dissector in the patent foramen caecum. F) CDS tract in contact with dura (white arrow). Case 3 (G-I), G) Patient's cutaneous stigmas: dimple, hypertrichosis and hemangioma. H) MRI in T1 sagittal sequence with contrast enhancement showing important intraspinal empyema. I) MRI in T1 axial sequence with contrast enhancement showing dermal sinus tract (white head arrow).

of patients complications such as meningitis, abscess, osteomyelitis, rupture of an associated epi/dermoid cyst, can be the first symptom, as we showed in the 3 cases presented, and even lead to permanent deficits and even death. Chances are higher in older ages (3,6,9,11). For that reason, once suspected, a further investigation should include ultrasonography (if in the lumbar region and before 3 months of age), CT-scan (thin slices) and/or MRI, as well a neurosurgical consultation for complete resection, the treatment of choice (7,8).

Cutaneous stigmas are a common presentation of occult dysraphism and fairly easily identifiable alterations on the physical examination of the pediatric population (3,5,6,8). Always in need of investigation, the positive predictive value rises when there is a combination of two or more signs. Early identification of dermal sinus potentially prevents complications and its consequent great morbidity and life-threatening risks (5,6,8). We presented a lumbar-sacral case with 3 cutaneous stigmas: a dimple, hypertrichosis, and a hemangioma, that was investigated only after having

severe complications, presented as a huge lumbar intradural empyema, and neurological deficits. It led to permanent deficits: paresis and neurogenic bladder, despite treatment. Singh et al (6), reported improvement of bowel/bladder function in only 1 of 8 patients.

In the 2 cranial cases presented, we were also able to show patients who were not diagnosed in the early examination due to a lack of investigation, only having the correct diagnosis and treatment after complications. In one series, 50% of the referred children presented with infection (4,8). Both of our cases were associated with a dermoid cyst.

Of cranial dermal sinuses, up to 85% are in the occipital region, as presented in case 1 (4). In this case, the ruptured dermoid cyst involved the brainstem, and the patient had to be submitted to a more aggressive surgery than if the diagnosis was made precociously. Although she had improvement of motor strength, ataxia persisted and the patient is now shunt-dependent.

Nasal dermal sinuses are extremely rare forms of CDS, with an estimated incidence of 1 per 40000 newborns (12), but once midline stigmas are seen, they should be investigated. Especially if there is hair protruding from a nasal pit, as in our case, because it is highly sensitive to dermal sinus. As in the case report of Hidalgo et al (10), the continuity of the skin to the dura was enabled by a patent foramen cecum. Another characteristic in imaging studies is the bifid crista galli (10,12–15). According to Pollock (13), the surgical approach chosen to excise the dermoid sinus and its tract should meet four criteria: access to all midline cysts; favor the rapid repair of cribriform defects; facilitate the reconstruction of the nasal dorsum; offer acceptable scar formation. The conventional approaches are based on a frontonasal bone flap in one or more steps (10,12,14). In our case, we modified this technique to a less aggressive approach, performing a low based bilateral frontal craniotomy and a small osteotomy around the tract for an “en bloc” resection. The bone was then returned to its original position and the periosteum, previously reflected, sutured in place, achieving the 4 criterias of Pollock, with good functional and aesthetic results.

Conclusion

The three presented cases illustrate the range of CDS presentation. Despite that, they all had one thing in common: late diagnosis leading to neurological deficits. Although well reported in the literature, CDS are usually diagnosed after complications. A detailed physical examination with thorough ectoscopy, especially in the midline should be performed in all newborns while actively searching for cutaneous stigmas of CDS. The knowledge of clinical presentation, early diagnosis and treatment are essential to prevent its life-threatening complications.

Disclosure Statement

The authors have no conflicts of interest to declare.

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