Congenital Stigmatic Alopecias Associated with Occult Spinal Dysraphism

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Received date: April 04, 2014; Accepted date: April 22, 2014; Published date: April 28, 2014

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Abstract

Spinal dysraphism is one of the most frequent malformations, and is defined as a spectrum of congenital anomalies characterized by an incomplete fusion of the midline mesenchymal, bony, or neural elements of the spine. Both the skin and nervous tissue anomalies which originate from ectoderm may occur simultaneously. It can be occult or cystic. Occult forms (occult spinal dysraphism) are characterized by skin-covered lesions without exposed neural tissue. The diagnosis of the occult spinal dysraphism is usually made based on a suspicion of the midline paraspinal skin lesions, mostly localized in the lumbosacral area such as subcutaneous lipomas, dermal sinuses, tails, sacral dimple, localized hypertrichosis, hyperpigmented lesions, hemangioma, mongolian spot, pigmentedary nevus, and aplasia cutis congenita. Here we described four male babies with occult spinal dysraphism associated with unusual alopecic scalp lesions.

Keywords: Alopecia; Congenital; Neonatal; Spinal dysraphism; Stigmata

Introduction

Occult spinal dysraphism (OSD) is described as a group of dystrophic conditions which occur under an intact dermis and epidermis [1]. Guggisberg et al. categorized patients with OSD according to the level of risk. 1-The high-risk group has one or more of the following:

A. Two or more congenital midline lumbosacral skin lesions of any kind.

B. Only one congenital midline cutaneous lesion associated with spinal cord dysfunction such as genitourinary and/or rectal problems or abnormal gait or abnormal arching or ulcers of the feet.

C. 1. Only one isolated but very suggestive congenital midline cutaneous lesion such as a lipoma, with or without an overlying PWS, dermal sinus, or tail.

2. The intermediate-risk group has isolated lesions such as aplasia cutis congenita, unclassified hamartoma, deviation in the gluteal furrow, and a typical dimple.

3. The last group includes patients with isolated lesions that are not usually associated with OSD, for example, PWS, hemangioma, hypertrichosis, simple dimple, pigmentedary nevus, and mongolian spot. These authors stated that a combination of two or more congenital midline lesions on the skin is a strong indicator of OSD [2].

Cases

Four male babies were admitted to our outpatient dermatology clinic due to their unusual alopecic scalp lesions. Patient 1 was admitted in April 2013, patients 2 and 3 were admitted in June 2013, and patient 4 was admitted in December 2013. Patients 1, 2 and 3 came from different cities of Turkey and they were Caucasian in origin, and patient 4 came from Syria, and he was Arabian in origin. None of the patients related to each other. Family histories of all the patients were unremarkable because no similar lesions or any congenital anomalies were reported. Each patient was the first child of their families. None of the parents of the patients had consanguinity, and all of the mothers denied any drug intake or infection before and during pregnancy. The parents of the patients indicated that their children were full-term babies and they were born through normal vaginal delivery.

The babies did not have any difficulties during their delivery, and their birth weights were normal. Patients 1, 2 and 3 were admitted to general practitioners several times due to both having skin lesions and crying very often. The doctors diagnosed them with urinary tract infections, and also stated that these scalp and skin lesions were unimportant. Patient 4 was diagnosed with gastro esophageal reflux and imperforate anus, at birth in Syria. Therefore, he was operated on and underwent colostomy, immediately after birth in his country. The parents of this patient also indicated that he had been diagnosed with recurrent urinary tract infection in the past. The parents of all the babies also stated that the babies were born with these bizarre alopecic lesions and have had no change in their appearances. In the dermatological examination of the 15-month-old child (Patient 1), there was a cross-shaped alopecic area on the occipital region with each line being 3.5x23 cm in size (Figures 1a and 1b). There was no additional scalp pathology such as in duration, scaling, erythema or open wound. In addition, a huge and symmetric mongolian spot that covered two thirds of the back (Figure 1c), a symmetric, 5x7 cm sized, dark-yellow triangular hyper pigmented plaque on the pubis (Figure 1d), and a blue nevus that was 1.2x1 cm in size on the left shoulder (Figure 1e) were seen.
In the systemic examination, a cryptorchidism and micro penis were detected. In the dermatological examination of the 4-month-old child (Patient 2), a big bilateral occipito-parietal regional alopecia was seen. There were hairs only on the temporo-parietal intersections, interparietal, and mid-frontal regions (Figures 2a,b,c). No other scalp pathology was found. In the sacral location 3.5 cm above the anus, a 0.8 mm in depth sacral dimple was detected (Figure 2d). In the systemic examination, a severe penile phimosis (Figure 2e), and three big mongolian spots on the back and buttocks were seen (Figure 2f).

Additionally, in the urological examination, we found a bilateral congenital hydronephrosis. In the dermatological examination of the 19 month-old child (Patient 3), an occipito-linear alopecic patch that was 17x3.5 cm in size was seen (Figure 3a). There was no other scalp pathology. In the sacral region 5 cm above the probable anal ostium, a sacral dimple was found (Figure 3b). Furthermore, bilateral costochondral hyperostosis and antero-lateral protrusions at the level of the tenth ribs were detected (Figure 3c).

The patient also had a pesequinovarus deformity. In the dermatological examination of the 10-month-old child (Patient 4), an oval 8x12 cm in size occipital alopecic patch was seen (Figure 4a). The patient did not have additional scalp pathology. In the sacral region approximately 7 cm above the probable anal ostium, a sacral dimple was found (Figure 4b). The patient had had a colostomy in the anal region. Additionally, perianal candidiasis and irritant contact dermatitis were seen around the colostomy (Figure 4c). In the genitourological examination, a penile phimosis and a cryptorchidism were detected (Figure 4d).

On the other hand, in the dermatoscopic examination of alopecic lesions of all the patients, the lesions were not truly alopecic but hypotrichotic. The count of the follicular orifices was equal in both the hairy and the alopecic areas. The thickness of the hairs decreased. A few terminal hairs were seen in the whole alopecic areas. No epidermal changing or scarring was seen on the hypotrichotic areas. Exclamation mark hairs were absent in the areas, and hair pull tests of the patients were also negative. Unfortunately, trichogram or scalp biopsy could not be performed because the parents of the patients did not give consent. In the Magnetic Resonance (MR) images of the central nervous systems of all the patients, there were openings between the vertebral arches. The openings were 4 mm at the level of L4-L5, 6 mm at the level of L5-S1, 4 mm at the level of L4-L5, and 5 and 6 mm at the levels of L4-L5 and L5-S1 respectively. The spinal cords of the patients were intact. The cranial MR images of the patients were normal. No neurological deficits were detected in the patients, and no additional neurological or other pathologies were found in the remaining clinical and laboratory examinations. According to our findings, the diagnoses of the patients were made as occult spinal dysraphism, and the alopecic lesions were also named “congenital stigmatic alopecias”.
Discussion

Both skin and nervous tissue are derived from the ectoderm. Spinal dysraphism (SD) refers to a spectrum of congenital anomalies characterized by an incomplete fusion of the midline mesenchymal, bony or neural elements of the spine. OSD is defined as skin-covered lesions without exposed neural tissue [2]. Spinal cord formation during embryogenesis consists of two stages: primary and secondary neurulation. Primary neurulation is characterized by the fusion of bilateral neural folds in the midline, and formation of the neural tube between the third and fifth gestation weeks. Secondary neurulation gives form to filum terminale and conus medullaris, and is characterized by the fusion and subsequent partial regression of caudal cell mass. Therefore, most of the spinal cords derived from primary neurulation and a part of the caudal tip of the spinal cord is derived from secondary neurulation [3]. On the other hand, it has been stated that spinal lipomas and tethered spinal cord were the most common spinal anomalies in OSD [2].

One of the findings of the SD is Congenital Dermal Sinus (CDS) and it is seen in about 2500-3000 live births. In the etiology of the CDS, it is usually accepted as separation failure between the neural and cutaneous ectoderm [3-5]. The sinus sometimes blindly ends in the subcutaneous tissue which is called dimple. [3,5]. Although Chern et al. (68%), and Kris and Desai (74%) showed that midline dimple was the most common skin lesion [67], only a typical dimple (≥5 mm in size, and ≥2.5 cm away from the anus) is highly associated with underlying SD [7]. Contrarily, Gibson et al. detected no pathology with ultrasonography in their 74 patients with dimple or pit, and they stated that these lesions alone do not indicate a high risk of OSD [8]. All 3 of our patients with dimples had atypical ones. Many cutaneous lesions associated with spinal dysraphism or OSD, have been reported, and these skin markers are important in order to recognize underlying spinal anomalies [2,6,9].

Besides the dermal sinus and dimple, other congenital midline paraspinal cutaneous lesions such as subcutaneous lipomas, tails, localized hypertichosis, hyper pigmented lesions, deviated gluteal furrow, pigmented nevus, fibroma pendulum and lumbosacral vascular lesions such as hemangioma or port-wine stain, mongolian spots and aplasia cutis congenitana can be found. Published documents have indicated that about 76% of patients with OSD have a single or combination of median or paramedian congenital lumbosacral cutaneous lesions [2]. Aside from the cutaneous and neurological findings, SD can be accompanied by urogenital and/or rectal problems such as recurrent infections or incontinence, ulcers or abnormal arch of the foot, abnormal gait [2], hydrenephrosis, nephrotic abnormalities [10], undescended testicle [11,12], micromenis [13], hypospadias [14], epispadias [15], tracheoesophageal fistula, imperforate anus, oesophageal atresia, omphalocoele, gastrochisis, cleft lip and palate, and orthopedic developmental disorders (pseudquinovaros, pesequinovalgos [12], hip dislocation [16-18].

In our patients, we detected six, five, four and five finding respectively, which can be attributed to SD. On the other hand, each patient with OSD also had different congenital and figured alopecia’s. According to our literature research, only two types of hair abnormalities of the scalp which are associated with underlying cranial neural tube defect or neurological sequelae have been reported [19-23]. One of them is “scalp hair tuft” and only a few cases have been reported up to now [19,24]. The lesion is described as a midline hair tuft over underlying rudimentary meningocele, and usually found on the occiput [19]. The other is “hair collar sign” that is collarette of dark, coarse hair around an circumscribed area of alopecia such as aplasia cutis congenita [22,25].

The sign is considered a relatively specific marker for cranial dysraphism [25]. None of the lesions of our patients were compatible with either of these two scalp lesions. Other probable causes of the non-scarring localized alopecia in neonates are temporal triangular alopecia (TTA) [26,27], and transient neonatal hair loss (TNHL) or neonatal occipital alopecia (NOA) [26,28]. TTA is a well-circumscribed triangular or lancet-shaped area of non-cicatricial hypotrichosis positioned in the fronto-temporal area [27,29]. The lesions are usually noticed in the second year of life when thevellus hairs are replaced by terminal ones. In the TTA, mostly vellus hairs are present in the affected area, and occasionally a few terminal hairs are retained. The hair density is normal, but the follicular size is abnormal and diminished [30,31].

The lesions of our patients were noticed by the parents at birth due to the fact that all babies were born with terminal hairs. However, because of their clinical dissimilarities to TTA, and the fact that the hair densities of the hypotrichotic patches decreased with the dermatoscopy, the diagnoses of TTA were excluded. On the other hand, non-marginal occipital alopecia [26], or neonatal occipital alopecia (NOA) is observed in the occipital area of infants after 8-12 weeks postnatally [28,32]. Its shape may be linear or oval [26,28]. Previously, the etiology of NOA has been thought to be friction caused by the neonate’s sleeping position. However, in recent years it has been considered that it might be due to physiological hair shedding [28,32], and it is thought that the condition is not an acquired alopecia, but a synchronized telogen effluvium after a prolonged anagen phase, which had begun in prenatal period. Another form of TNHL is marginal, often band-like alopecia of the fronto-temporal region of the scalp [26,28,32].

It may be confused with alopecia areata, but in the alopecia areata, hair pull-test is always positive [26]. Recently, Neri et al. have proposed a new way to classify TNHL: 1-Neonatal type is rare and appears in the first 4 weeks of life with a fronto-temporal pattern. 2-Classic type is more common than the first and appears in 8-12 weeks of life with a predominant occipital pattern [33]. Additionally, we had previously presented a patient who had a combination of the two forms of NOA or TNHL. This patient had both the occipito-linear alopecia and bilateral triangular fronto-temporo-parietal alopecic patches. Therefore, we suggested that the combined form might be a third and different clinical type of TNHL, and the triangular patches might be a triangular variant of the marginal form [34]. The cross-shaped lesion of our first patient did not resemble any alopecia described in previous literature. On the other hand, the lesions of the second patient resembled an enlargement of the combined form of TNHL. Also, the lesions of the last two patients were consistent with the non-marginal forms (occipital-linear, and occipital-ovale) of the TNHL clinically. We could not do any scalp biopsies due to the fact that the parents of the patients did not give consent. However, the diagnoses of the previously reported cases of TNHL have only been made based on clinical and dermatoscopic findings [28,32,33].

Although the shapes of the lesions of each patient were different, because all the hypotrichotic patches usually have thinned hairs similar to TNHL, we first thought these lesions (especially the lesions of the first and second patient) might be new and different clinical forms of the TNHL. On the other hand, TNHL is a self-limited and transient disease, and there is no need for treatment [28,32]. However, because our patients were very young, we do not know how the shapes
of the alopecic lesions will look in the future. None of the published OSD reports included information about these unusual accompanying alopecias like the ones found in our patients. Therefore, even though these alopecias resembled TNHL morphologically, it was thought the underlying congenital defects, which cause these spinal dysraphisms, might have created the alopecic lesions. Also, these different clinical manifestations of the alopecias accompanying OSD might have formed depending on the site, evaluation stage, and severity of the dysraphisms in utero. To the best of our knowledge, the alopecias we presented are the first reported cases in literature in terms of accompanying OSD, and they are likely to be stigmatic scalp lesions of OSD. However, even though it was not previously reported, a probable connection between TNHL and OSD may be found in the future studies. On the other hand, the skin findings such as tails have not been well defined by physicians and usually treated surgically, forpurposely esthetic reasons without any investigation of underlying OSD [2].

Therefore, a careful neurological examination of any patient with similar findings to our patients especially of unusual alopecia, sacral dimple, giant mongolian spot, symmetric pubic pigmentation, penile phymosis, microgenital, congenital hydronephrosis, imperforate anus, cryptorchidism, and rib anomalies must be carried out. Guggisberg et al. indicated that an MRI must be performed even if previous ultrasonography (USG) findings were normal for the high-risk group. In addition, this examination for the intermediate-risk group is necessary for children older than 6 months, but optional for children younger than 6 months with doubtful or abnormal USG findings. Lastly, it is not necessary for the low-risk group [2]. Although our patients were in the different risk groups (low for patient 1, intermediate for patients 2 and 3, and high for patient 4), an OSD was detected in each of their MRI. Therefore, we suggest that in every suspected case of OSD, an MRI should be performed regardless of the risk-group. USG which is an inexpensive, quick and noninvasive screening method for OSD and it can be applied easily. But, it is more useful for neonates under the age of 5 to 6 months because the posterior elements of the spine are non ossified [2].

In conclusion, when faced with a neonatal, symmetric and unusual stigmatic alopecia, careful attention must be paid to underlying congenital midline anomalies, especially OSD. On the other hand, both specialists including dermatologist, and general practitioners should be aware of any unusual alopecias and the other cutaneous findings of OSD. To diagnose the probable underlying anomalies early, imaging studies must be performed to evaluate in suspected cases, without delay. Hence, redundant interventions can be avoided, and their potential path physiological and psychopathological effects can be prevented. Additionally, in such cases, a genetic counseling of the family regarding the probable risk of recurrence should be recommended.

References


