Rhabdomyomatous Mesenchymal Hamartoma Associated with Congenital Anomalies: Report of an Unusual Perineal Case

Jia-Ru Wang  Tseng-Tong Kuo  Ke-Jen Yu  Po-Yu Shih  Yue-Zon Kuan  Chih-Hsun Yang

The ectopic occurrence of skeletal muscles in skin is an unusual condition. These lesions are named rhabdomyomatous mesenchymal hamartoma now. More accurately speaking, this entity should be called “cutaneous rhabdomyomatous choristoma” because the skeletal muscle is not a normal element in skin. We described a case occurring in perineal region associated with amniotic band syndrome, imperforate anus, and intestinal atresia. The association with gastrointestinal tract anomaly has not been reported previously. This unusual case is documented and compared to two other cases reported in this region. (Dermatol Sinica 26: 93-98, 2008)

Key words: Rhabdomyomatous, Hamartoma, Choristoma

INTRODUCTION

The ectopic occurrence of skeletal muscles in skin is an unusual condition. Thirty-three cases have been reported in the literature under various names of hamartoma. We saw a case occurring in perineal region. This unusual case is documented and compared to two other reported cases occurring in perineal region.

CASE REPORT

A full-term Taiwanese male baby was born to a G2P2 mother with normal birth weight (3850 gm). Regular antepartum examinations were uneventful and did not reveal any abnormalities. She denied any invasive antepartum procedures, including amniocentesis. Multiple congenital anomalies, including high type imperforate anus with recto-bulbar urethral fistula, atresia of large intestine and rectum, hypospadia, scrotal bifida with severe chordee, and anomalies of the right big and little toes (amniotic band syndrome), were noted at birth. Multiple irregular, confluent hypopigmented macules and plaques and several small satellite lesions were found in the lower part of the scrotum, peri-anal area left to the imperforate anal dimple, and bilateral coccygeal regions (Fig. 1). The clinical differential diagnosis included nevus anemicus, nevus depigmentosa, segmental vitiligo and lichen sclerosis et atrophicus. A skin biopsy specimen, which was taken from the skin 2 cm away from the left side of the imperforate anal dimple, revealed mild acanthosis with decreased pigmentation of the basal layer and multiple skeletal muscle fibers in dermis and subcutis (Fig. 2A). Cross striations were evident in these skeletal muscle fibers (Fig. 2B). Pilosebaceous structures and sweat glands appeared to be normal. There were loose fibrous bands in subcutis and the fat cells showed variation in sizes (Fig. 2C). Immunostaining for HHF-
Rhabdomyomatous Mesenchymal Hamartoma 35 (muscle actin antibody, Dako, Denmark), MyoD1 (Myogenin D1, Dako, Denmark) and Desmin (Dako, Denmark) confirmed the ectopic presence of striated muscle fibers (Fig. 2D).

Ultrasonographic examination of the brain via the anterior fontanel revealed normal midline structure, including corpus callosum. Chromosome study demonstrated normal karyotype (46, XY). Trisomy was therefore excluded. Atresia of large intestine and rectum, imperforate anus, hypospadia and scrotal bifida were corrected in sequential surgeries. Hirschsprung’s disease, a possible etiology of intestinal atresia, was excluded due to the presence of ganglion cells in the anorectal tissue. The infant was physically well at age of 5 months old.

DISCUSSION

By definition, hamartomas are benign tumor-like nodules composed of overgrowths of mature cells and tissues, but with disorganization and often with one predominant element. In the skin, aberrant proliferations of several cellular lineages, including smooth muscle, sweat glandular, pilus, fibroelastic, vascular, neural, and melanocytic elements can occur. Choristoma is defined as a mass formed by the faulty development of tissue not normally found at that site.

The pathologic characteristics of the present case are the same with the findings of the reported cases of rhabdomyomatous mesenchymal hamartomas. The entity was first described in 1986 by Hendrick et al. as “striated muscle hamartoma” and subsequently renamed “rhabdomyomatous mesenchymal hamartoma” (RMH) or “congenital midline hamartoma.” More accurately speaking, this entity should be called “cutaneous rhabdomyomatous choristoma”, because the skeletal muscle is not a normal element in skin. The clinical manifestations are variable, including pedunculated polyps, subcutaneous nodules, or sessile masses and papules. To our knowledge, there are only 33 reported cases in the English language literature. Of these, there is a 1.35:1 male-to-female preponderance. It occurs most commonly in the head and neck regions where striated muscles, such as the orbicularis oris, platysma, orbicularis oculi, are superficially located, suggesting aberrant embryonic migration of striated muscle fibers into the dermis as a possible etiology.

Including the present patient, only 3 cases occurring in the perineal regions have been reported. (Table 1) The anal canal, like the head and neck, is adjacent to developing skeletal muscles during embryogenesis, allowing potentially aberrant migration of rhabdomyogenic elements into superficial locations during development. The first report of a lesion arising in perianal region was in a 7-month-old girl associated with a hemangioma in the same area that later regressed completely. The second patient, a 3-month-old girl, presented with a polypoid lesion in which disorganized arrays of skeletal muscle fibers with admixed adipose tissue and smooth muscles in dermis. No other congenital anomaly was mentioned in either case.

Rhabdomyomatous mesenchymal hama-
Rhabdomyomatous Mesenchymal Hamartoma may present sporadically as a solitary lesion in a normal neonate as in the majority of cases or can be associated with multiple congenital anomalies, including amniotic band syndrome, ocular abnormalities, thyroglossal duct sinus and Delleman’s syndrome. Delleman’s syndrome consists of colobomas, an absent corpus callosum, orbital cysts, proencephalic cysts, and facial skin tags. The normal midline structure, including corpus callosum, and the absence of facial skin tags in the present case exclude the possibility of Delleman’s syndrome.

The amniotic band syndrome is a collection of fetal malformations associated with fibrous bands that appear to entangle or entrap various fetal parts in utero, leading to deformation, malformation, or disruption. The etiology is unknown. Hendrick et al. postulated that traction on the skin by amniotic adhesions may predispose toward the development of a rhabdomyomatous mesenchymal hamartoma. There have also been reports associating amniotic band syndrome with amniocentesis. However, the mother of our patient did not receive amniocentesis during antepartum examination.

In addition to amniotic band syndrome, our patient also had imperforate anus, intesti-
nal atresia, hypospadia, and scrotal bifida with severe chordee. Anorectal malformations occur in 1 in 4000 to 1 in 5000 newborns and are more common among males and in children with Down syndrome (Trisomy 21). Anal malformations are believed to result from failure of migration of the anus and excessive fusion. The higher and more complex the anorectal malformations, the greater the chance of a life-threatening defect, especially a urologic anomaly. Urogenital defects, such as hypospadias, vesicoureteral reflux, cryptorchidism, rotated kidney, neurogenic bladder and renal agenesis occur in 48% of patients with anorectal malformations. Atresia of large intestine and rectum, imperforate anus with recto-bulbar urethral fistula, hypospadias and scrotal bifida were corrected in sequential operations by pediatric surgeons in the present case. The association of rhabdomyomatous mesenchymal hamartoma with gastrointestinal tract anomalies has not been reported previously. Although rhabdomyomatous mesenchymal hamartoma may present as a solitary lesion in a neonate in most cases, systemic evaluation for other congenital anomalies is warranted because of possible common etiologies.

In summary, we report a male neonate presenting with rhabdomyomatous mesenchymal hamartoma of the perineal region associated with amniotic band syndrome, imperforate anus and intestinal atresia. The association with gastrointestinal and genitourinary tract anomalies has not been reported previously. Although rhabdomyomatous mesenchymal hamartoma may present as a solitary lesion in a neonate in most cases, systemic evaluation for other congenital anomalies is warranted because of possible common etiologies.

**REFERENCES**


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Table 1: Comparison of Features of Reported Perineal Cases

<table>
<thead>
<tr>
<th>Case no. (reference)</th>
<th>Age/sex</th>
<th>Clinical presentation</th>
<th>Microscopic findings</th>
<th>Congenital anomalies</th>
<th>Appellation</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>7 month/F</td>
<td>Perineal hemangioma appear 15 day after birth, hemangioma regressed leaving polypoid lesion</td>
<td>Mature skeletal muscle fibers admixed with adipose tissue in dermis</td>
<td>None reported</td>
<td>Striated muscle hamartoma</td>
</tr>
<tr>
<td>2</td>
<td>3 month/F</td>
<td>A papillary lesion in the perianal area</td>
<td>Mature skeletal muscle fibers with admixed adipose tissue and smooth muscles in deep dermis</td>
<td>None reported</td>
<td>Cutaneous mesenchymal hamartoma with mixed myogenous differentiation</td>
</tr>
<tr>
<td>3 (Present case)</td>
<td>1 day/M</td>
<td>Hypopigmented macules with several small satellite lesions in the lower part of the scrotum, left peri-anal dimple, and bilateral coccygeal regions</td>
<td>Hypomelanosis, skeletal muscles in upper and lower dermis and subcutis with lipoatrophy</td>
<td>Imperforate anus, atresia of large intestine, hypospadia, scrotal bifida, and amniotic band syndrome</td>
<td>Cutaneous rhabdomyomatous choristoma</td>
</tr>
</tbody>
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Dermatol Sinica, Jun 2008
Rhabdomyomatous Mesenchymal Hamartoma

橫紋肌間葉型錯構瘤伴隨先天異常：一個罕見的會
陰部病例報告

王佳茹  郭承統  游可任  石博宇  官裕宗  楊志勛
台北長庚醫院皮膚科  病理科  長庚醫學大學醫學系

橫紋肌異常的在皮膚出現是一個稀有的狀況。這樣的病灶目前被命名為橫紋肌間葉型錯
構瘤，但因為橫紋肌並不屬於正常皮膚的成分，更正確的來說，他應該被命名為皮膚橫紋肌
迷離瘤。我們提出一個發生在會陰部的病例，他同時合併了羊水袋症候群、無肛症及腸道閉
鎖。伴隨著先天性腸胃道異常的情況以往並未曾被報告過。我們報告一個罕見的病例並且和
另外兩個發生在會陰部的病例作比較。（中華皮誌：26: 93-98, 2008）