Infantile myofibromatosis - diagnosis and treatment in the neonatal period

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This article was submitted to RAD CASA - Medical Sciences as the original article

Conflict of Interest Statement: The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Received: 5 October 2021
Accepted: 15 November 2021
Published: 27 December 2021

Citation:
Vrdoljak V, Tumbri J, Paleka Bosak E, Brletić D, Radina Jurčić S, Gverić-Ahmetašević S. Infantile myofibromatosis - diagnosis and treatment in the neonatal period
RAD CASA - Medical Sciences. 548=56-57 (2021): 116-118
DOI: 10.21857/90836c7qdy

Abstract:

Infantile myofibromas are benign congenital soft tissue tumors that may be present at birth or present later during the infant period. They are most often solitary, but can also be multicentric or generalized, and therefore additional radiological processing is required in order to exclude the generalized form. The treatment method is surgical excision followed by pathohistological confirmation. This article describes a case of infantile myofibroma in a term female newborn diagnosed immediately after birth.

Keywords:

infantile myofibromatosis, benign skin tumor, children, surgical excision

Sažetak:

Infantilna miofibromatoza – dijagnoza i liječenje u neonatalnom razdoblju

Infantilni miofibromi su benignu urođeni tumori mekog tkiva koji mogu biti prisutni po rođenju ili se prezentirati kasnije tijekom dojenačkog razdoblja. Najčešće su solitarni, ali mogu biti i multicentrični ili generalizirani te je stoga potrebna dodatna radiološka obrada u svrhu isključenja generalizirane forme. Metoda liječenja je kirurška ekscizija nakon čega slijedi patohistološka potvrda. U ovom članku opisan je prikaz slučaja infantilnog miofibroma kod terminskog ženskog novorođenečeta koji je dijagnosticiran neposredno nakon poroda.

Ključne riječi:

infantilna miofibromatoza, benigni tumor kože, djeca, kirurška ekscizija

Case Report

In this article, we present a female term newborn, born from a normal course of pregnancy who was observed to have a pedunculated skin lesion on the fifth finger of her left hand immediately after delivery (Figures 1 and 2). On the fifth finger of the left hand, a cystic formation is observed within which there is a multilobulated tumor formation. Infantile myofibroma has been suspected. The newborn was of orderly other clinical status. Surgical excision of the lesion was performed and the lesion was sent for pathohistological evaluation. Pathohistological findings confirmed the clinical diagnosis of infantile myofibroma. An ultrasound of the abdomen and solitary organs was performed, as well as an ultrasound of the brain, which was in order. No myofibroma was reported in parents and relatives. During the follow-up period of the next 6 months, no new lesions appeared.

Figure 1.

Figure 2.

Discussion

Infantile myofibroma is the most common fibrous tumor of infancy (1). In 60 percent of cases, they are congenital, but may occur later in infancy. (2-4). They appear more often in male children. The etiology of the disease is not completely clear. In most cases, they are sporadic, but intra-family occurrences have been reported. Autosomal recessive as well as autosomal dominant inheritance patterns have been described (5-7). A mutation in the PDGFRB gene (platelet-derived growth factor receptor-beta) has been identified as the causative agent of an autosomal dominant form of the disease (8,9). The same mutation has been described in some cases of a sporadic form of the disease (10,11). Also, mutations in the NOTCH3 gene have been described within some familial forms (12). The clinical presentation is heterogeneous, but infantile myofibromas are mainly presented as solid masses, purple or flesh-colored, or as subcutaneous masses covered with normal skin appearance. The size ranges from several millimeters to several centimeters, and the surface can be regular, lobulated, or ulcerated (13). They can appear as a solitary or as a multicenter and generalized form. The most common is a solitary form that affects the dermis, subcutis, but can also affect deeper tissues and bone. In solitary lesions, the head and neck are usually located, followed by the upper extremities (1, 13,14). In the multicentric form, visceral organs can be affected, and an important manifestation is spinal involvement, which may end with lethal outcome. The generalized form includes involvement of the visceral organs; lungs, gastrointestinal system, and central nervous system and as such has the highest mortality (15). Less commonly, myofibromas occur in the form of pedunculated lesions with a multilobular surface. Treatment includes surgical excision, and the diagnosis should be confirmed by pathohistological findings. A characteristic pathohistological finding includes a peripheral zone in which spindle cells are arranged in intertwined bundles and a central zone with more rounded cells with a perivascular arrangement, extending from the dermis to the subcutaneous tissue.

Conclusion

Infantile myofibroma is a benign skin tumor of infancy that may be present at birth or appear later in infancy. The prognosis is generally good, unless it is a generalized form. Treatment involves complete surgical excision of the lesion after which the diagnosis should be confirmed by pathohistological findings.

Author Contributions:

All authors listed have made substencial and intelectual contribution to the work, and approved it for publication.

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