A RARE ASSOCIATION BETWEEN GIANT CONGENITAL MELANOCYTIC NEVUS AND UNILATERAL ANOPHTHALMIA

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Summary

Giant congenital melanocytic nevus is a melanocytic lesion present at birth that reaches a diameter greater than 20 cm and occurs in 1 per 500,000 newborns. Melanocytes are found in utero at about 40 days of gestation and it is thought that congenital nevi develop between the 2nd and 6th month of gestation. The orbital region is a rare site of occurrence of congenital hairy melanocytic nevus, especially the giant form. We present a rare case of a giant congenital nevus that involves the head and neck with congenital unilateral anophthalmia in a one day old girl, coming from an uninvestigated pregnancy from a 25 year old multipara woman.

***Keywords**: giant congenital nevus, anophthalmia, dermatoscopy, immunohistochemistry.*

Received: 29.01.2021 *Accepted: 5.03.2021*

Introduction

Giant congenital melanocytic nevus is a melanocytic lesion present at birth that reaches a diameter greater than 20 cm and occurs in 1 per 500,000 newborns [1]. Melanocytes are found *in utero* at about 40 days of gestation and it is thought that congenital nevi develop between the 2nd and 6th month of gestation [2]. The orbital region is a rare site of occurrence of congenital hairy melanocytic nevus, especially the giant form. Patients with congenital melanocytic nevus have long been known to be at risk of melanoma. Regarding giant nevi, 50% of the malignancies develop by 3 years old, 60% by childhood, and 70% by puberty [3]. Congenital anophthalmia is rare and may cause deficient orbitofacial growth and impaired visual capability. Possible risk factors are: disturbances of the morphogenetic pathway, maternal and gestational variables, patient characteristics, genetic alterations and environmental factors or drugs [4].

Case presentation

We present a rare case of a giant congenital nevus that involves the head and neck with congenital unilateral anophthalmia in a one day old girl, coming from an uninvestigated pregnancy from a 25 year old multipara woman. The giant congenital nevus has 21,3 cm diameter with rough surface and lackluster hairs (fig. 1).

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Figure 1. Giant congenital melanocytic nevus with anophthalmia

Other examinations and surveys were normal. At birth, the girl had an Apgar score of 9, 2690 g, 49 cm long and no other malformations. No pregnancy-related trauma was reported. There was no family history of melanocytic lesions and the parents along with their other 3 children were healthy. No underlying abnormalities of the skull or intracranial structures were noted on ultrasound examination.

Discussions

Dermoscopy revealed multiple islands of color and irregular pattern. Giant congenital nevi are often heterogenous due to multiple patterns within the same lesion, but each island tends to be fairly homogenous in its appearance [5]. The nevus presented reticular network, brown-black globules in a cobblestone pattern, diffuse homogenous pigmentation and terminal hair.

The biopsy confirmed the diagnostic of congenital melanocytic nevus. Microscopic examination revealed a fragment with connective-adipose tissue which presents melanocytes proliferation with oval, small and monomorphic nuclei and a small amount of eosinophilic cytoplasm. Regarding anophthalmia, histopathological evaluation of orbital contents usually reveals an extremely small or malformed globe with only rudimentary ocular contents. Overall, extraocular muscles are often absent or markedly decreased in anophthalmia.

The immunohistochemical test has shown a diffuse cell proliferation, intense positive for S100, melan-A, Mitf and SOX10, reaffirming the melanocytic nature of the lesion. The cell proliferation is also positive for vimentin and some cells are also positive for CD68. The proliferation index ki67 is 6-8%. The cell

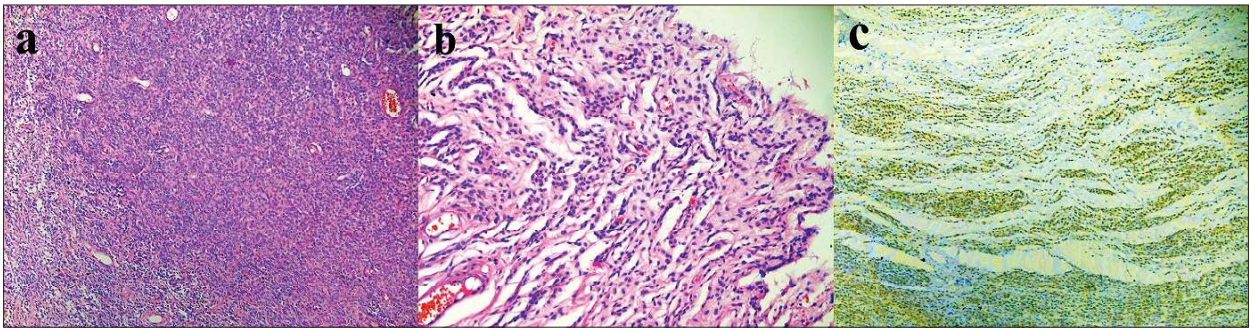

Figure 2. a) haematoxylin eosin staining (100x) b) haematoxylin eosin staining (200x) c) immunohistochemical staining MelanA (100x)

proliferation is negative for CD1a, desmin, actin, CD34, Mic2 and CLA (fig.2).

Giant congenital nevi involving the head or neck, may associate meningeal melanocytosis and they may be complicated by seizures, focal neurologic defects, obstructive hydrocephalus or malignant changes. The MRI should be performed when the patient is 4-6 months old, therefore the girl will be investigated accordingly [6].

Giant congenital nevi are associated with cutaneous melanoma, leptomeningeal melanoma, neurocutaneous melanocytosis and malformations of the brain, therefore the baby will be monitored long term, due to the high risk [7].

Anophthalmia occurs when the neuroectoderm of the primary optic vesicle fails to develop properly from the anterior neural plate of the neural tube during embryological development. European rates are similar to those seen in the United States and have been reported as 0.19 case per 10,000 births [8]. Recent studies have shown an elevated prevalence in some ethnic groups. These groups include Pakistani and Scottish children. Genetic, environmental, and possible classification issues may explain these high rates of disease and need to be explored further [9].

Physicians giving bad news have to choose their words with maximum responsibility and in this case maintaining hope and psychological mobilization of the parents are essential to the subsequent cooperation of the medical professionals with the patient's parents [10].

Conclusions

The management of congenital melanocytic nevi depends on a number of factors, including the size and the location of the lesion, patient's age, the effect on cosmesis and the potential for malignant transformation. The treatment of giant congenital nevi depends on the potential of malignant transformation and the cosmetic appearance, therefore follow up was recommended [11]. In addition, we emphasize the importance of following up a pregnancy to prevent the abnormalities that can occur.

 # All authors have equally contributed to this paper.

Bibliography

1. Castilla EE, da Graça Dutra M, Orioli-Parreiras IM. Epidemiology of congenital pigmented naevi: Incidence rates and relative frequencies. *Br J Dermatol* 1981, 104(3):307-15
2. Ghulam SH, Syed SA, Shahla K. Congenital giant melanocytic nevi. *Rare Tumors* 2009, 1(1):e9
3. Kinsler VA, O'Hare P, Bulstrode N, Calonje JE, Chong WK, Hargrave D, Jacques T, Lomas D, Sebire NJ, Slater O. Melanoma in congenital melanocytic nevi. *Br J Dermatol* 2017, 176(5):1131-1143
4. Llorente-Gonzales S, Peralta-Calvo J, Abelairas-Gómez JM. Congenital anophthalmia and microphthalmia: epidemiology and orbitofacial rehabilitation. *Clin Ophthalmol* 2011, 5:1759-1765
5. Seidenari S, Pellacani G. Surface microscopy features of congenital nevi. *Clinics in Dermatology* 2002, 20(3):263-267.
6. Jellinger K, Bock H, Brenner H. Meningeal melanocytoma: report of a case and review of the literature. *Acta Neurochir* 1988, 94:78-87.
7. Kaplan EN. The risk of malignancy in large congenital nevi, Plast Reconstr Surg 1974, 53(4):421-8.
8. Boyd PA, Haeusler M, Barisic I. EUROCAT Report 9: Surveillance of congenital anomalies in Europe 1980-2008. *Birth Defects Res A Clin Mol Teratol*. 2011, 1991 Suppl 1:S1.
9. Shah SP, Taylor AE, Sowden JC, Ragge NK, Russell-Eggitt I, Rahi JS. Anophthalmos, microphthalmos, and typical coloboma in the United Kingdom: a prospective study of incidence and risk. *Invest Ophthalmol Vis Sci*. 2011, 52(1):558-64.
10. Constantin DA, Cioriceanu IH, Tantu MM, Popa D, Badau D, Burtea V, Nemet GC, Rogozea LM. Ethical dilemmas in communicating bad news following histopathology examination *Rom J Morphol Embryol* 2017, 58(3):1121-1125.

11. Gheucă-Solovastru L, Vâță D, Stătescu L, Constantin MM, Andrese E. Skin cancer between myth and reality, yet ethically constrained. *Rom J Bioet*. 2014, 12(2):47-52.

Conflict of interest
NONE DECLARED

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