#22

WILHELMSTIFT
Katholisches Kinderkrankenhaus

Belgian Dermatology Days 2024
21 & 22 MARCH 2024
BMCC, Bruges

Congenital plaque-type elastoma: a new morphological entity

Braeckmans D[1], Rütten A[2], Hoeger PH[3]
[1]GZA Hospitals, Antwerp (Belgium)
[2] Institute for Dermatopathology, Friedrichshafen/Bodensee (Germany)
[3]Department of Paediatric Dermatology, Cath. Children's Hospital Wilhelmstift, Hamburg (Germany)

Background

Elastomas are rare, clinically asymptomatic, skin coloured, elastin-rich connective tissue naevi. They present at birth or during infancy either as solitary or disseminated (Buschke-Ollendorff syndrome) lesions. Here we describe three cases of a non-syndromal, congenital, plaque-type elastoma in the neck area which to our knowledge has not been described before.

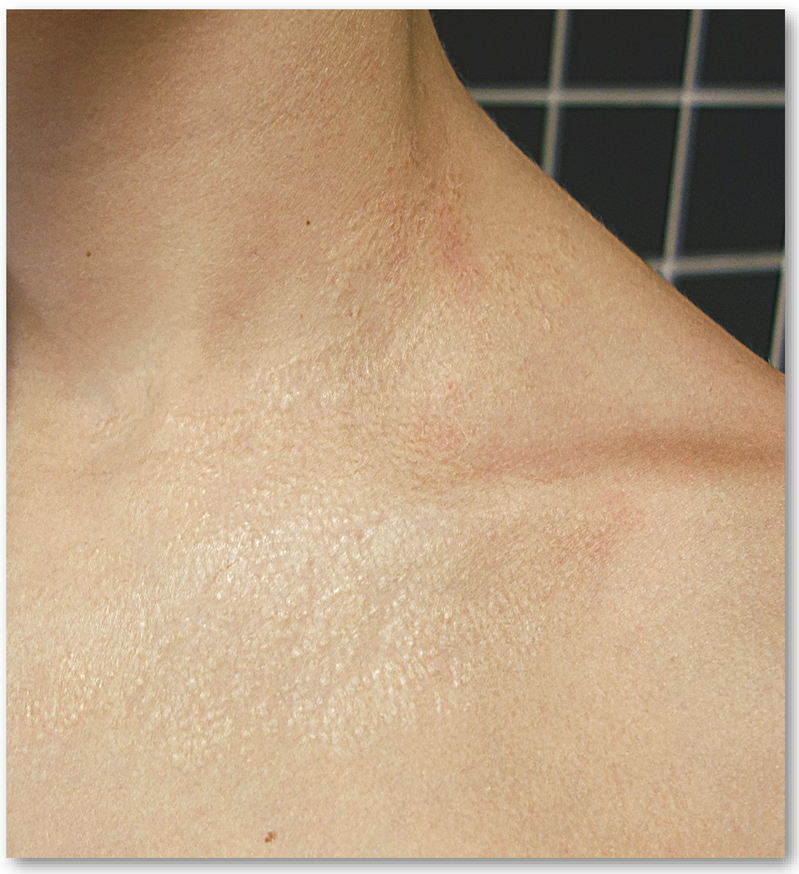
Case report

We describe three cases of elastomas presenting as congenital plaques in the neck area. All three patients presented to our out-patient department at the age of 4-17 months with an asymptomatic, skin-coloured or slightly yellowish plaque (diameter: 6-9 cm) on the upper chest/neck region, not accompanied by skin laxity. The plaques were asymptomatic. Family history was negative for similar lesions. There was no evidence of cardiovascular or ophthalmological disease.

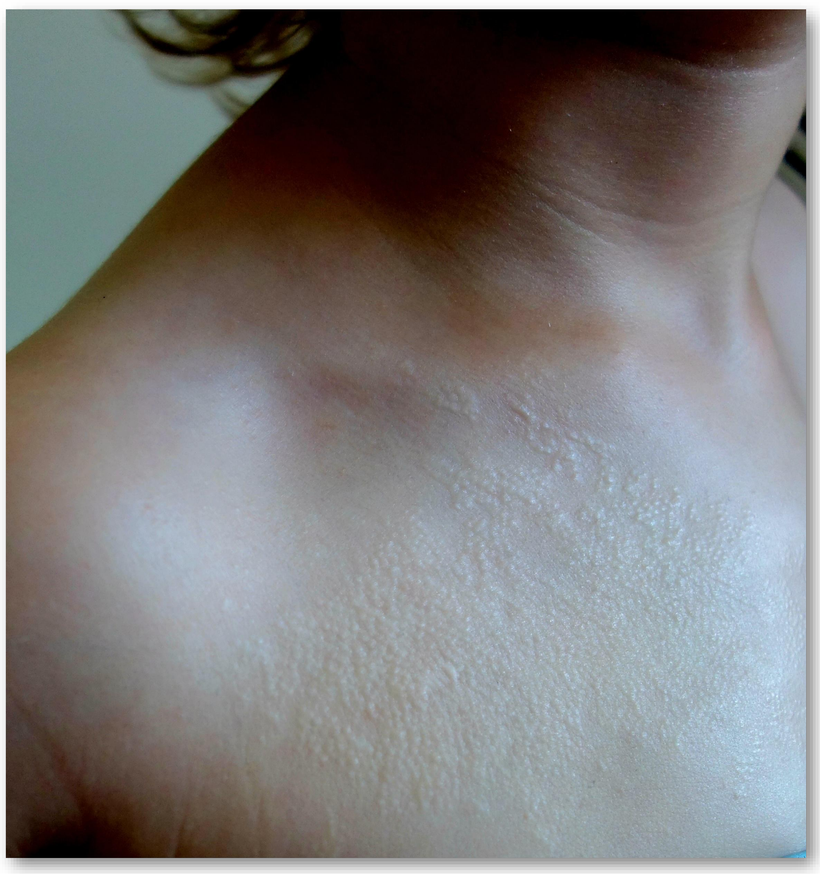
Histopathology

We recommended histopathological examination in order to exclude differential diagnoses such as pseudoxanthoma elasticum. Elastic stain revealed an increased number of horizontally arranged small normal looking elastic fibres, in accordance with our clinical diagnosis of elastoma.

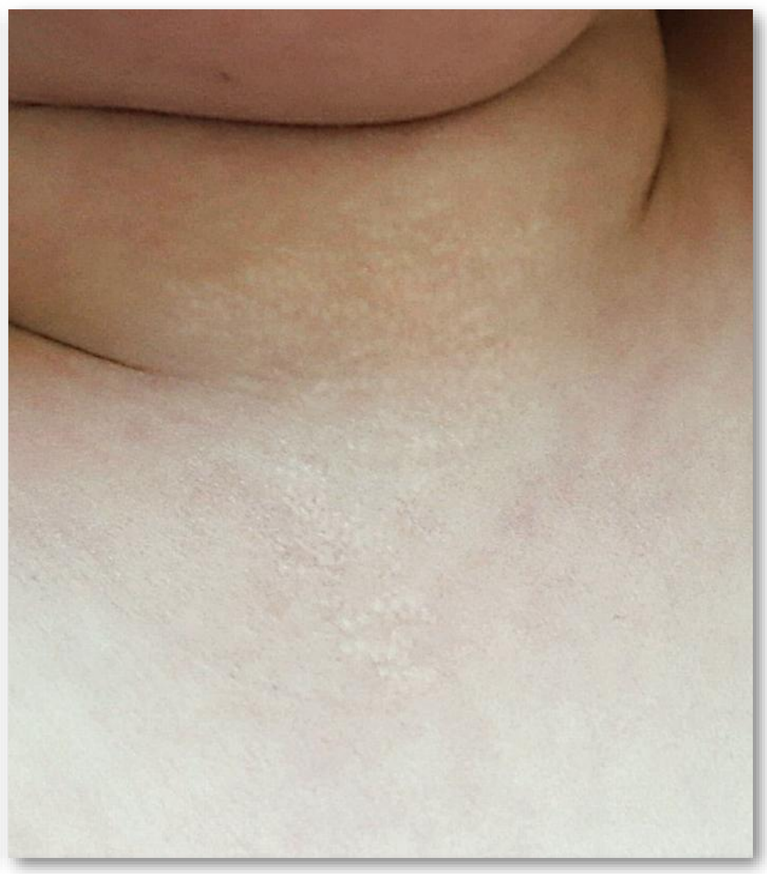
Case no.	Age at manifestation [months]	Age at presentation [months]	Localization	Size at presentation [cm]	Skin laxity
1	birth	4	upper chest/ anterior neck	7 x 3	no
2	12	17	upper chest	8 x 9	no
3	birth	14	upper chest/ anterior neck	6 x 4	no



Case no. 1 (age 9)



Case no. 2 (age 2)



Case no . 3 (age 1)

Discussion

To the best of our knowledge, no similar plaque-type elastomas have been reported before, making these unique morphological distributions of a well-known but rare connective tissue naevus. Their clinical similarity to Buschke-Ollendorff syndrome (BOS) associated connective tissue naevi and pseudoxanthoma elasticum, respectively, warrants histological evaluation which is able to separate these entities. In BOS, a rare autosomal dominant trait caused by a loss-of-function germline mutation of *LEMBD3*, juvenile elastomas are associated with osteopoikilosis, a skeletal dysplasia featuring multiple dense trabecular foci. Pseudoxanthoma elasticum (PXE) is an autosomal recessive disorder, caused by mutations in the *ABCC6* transporter gene, leading to calcification and fragmentation of the elastic tissue of the skin and extracutaneous organs (retina, arteries, gastrointestinal tract). This may lead to vision loss, cardiovascular disorders or gastrointestinal bleeding. Because of its potential life threating effects, PXE is a diagnosis that should not be overlooked.

Treatment of elastomas is usually not required and we did not recommend surgical removal in our patients due to the benign nature of elastoma and the resultant, sizeable scar. Standard follow-ups after histopathological validation of the diagnosis are not indicated.

Literature

Lewandowsky F. Über einen eigentümlichen Naevus der Brustgegend. Arch Derm Syph 1921; 131:90-94
Buschke A, Ollendorff H. Ein Fall von Dermatofibrosis lenticularis disseminata. Derm Wochenschr 1928; 86:257–262
Hellemans J, Preobrazhenska O, Willaert A et al. Loss-of-function mutations in LEMD3 result in osteopoikilo-sis, Buschke-Ollendorff syndrome and melorheostosis. NatGenet 2004; 36:1213–1218
Pope V, Dupuis L, Kannu P et al. Buschke-Ollendorff syndrome: a novel case series and systematic review. Br J Dermatol. 2016; 174:723.
Saussine A, Marrou K, Delanoé P et al. Connective tissue nevi: an entity revisited. J Am Acad Dermatol. 2012; 67:233-9
Bercovitch L, Patrick T. Pseudoxanthoma elasticum. J Am Acad Dermatol. 2004; 51: S13–S14.
Socrier Y, Wann AR, Sigal ML et al. Syndrome de Buschke-Ollendorff. Ann Dermat Vénéréol 2014; 141:164-166.
Lewis GK et al. Acquired disorders of elastic tissue: Part I. Increased elastic tissue and solar elastotic syndromes. J Am Acad Dermatol 2004; 51: 1-21

Delphine Braeckmans
delphine.braeckmans@zas.be