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Case Report
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Vascular Transformation of Lymph Node Sinuses and Williams Syndrome: A Case Report

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Background

Vascular Transformation of lymph node Sinuses (VTS) is a rare, acquired and benign process in which the sinusoidal architecture of lymph nodes is replaced by an anastomosing network of vascular channels ranging from capillaries to cavernous spaces, without involving parenchyma, capsule, or perinodal fibroadipose tissues. Often it is presented as enlarged lymphadenopathies. The postulated mechanism is intussusceptive lymphangiogenesis (with the development of pillars, interstitial tissue structures, folds, vessel loops and septa), and it has been associated with efferent lymphatic and/or venous obstruction, due to tumor, thrombus, or as a result of surgery or radiotherapy. However, angiogenic factors produced locally by activated lymphoid cells seem to play a role, and can also produce VTS without lymphovascular obstruction. The diagnosis is histological.

Williams syndrome is a rare genetic and neurodevelopmental disorder, with autosomal dominant transmission, due to deletion at the chromosome band 7q11.23, that involves the elastin gene. After birth, infants often present with failure to thrive, short stature and supra-vascular aortic stenosis. Children also may have other elastin arteriopathies, due to stenosis of medium and large arteries owing to thickening of the vascular media from smooth muscle, as peripheral pulmonary stenosis, hypertension, mitral valve insufficiency and renal artery stenosis. Often, they have endocrine abnormalities, connective tissue abnormalities, global cognitive impairment and psychiatric disorder [1-3]. We describe a singular case of association of VTS and Williams syndrome.

Case Presentation

A 24-year-old man presented to our outpatient consultation with a 2-month history of right axillary swelling, growing progressively. He was diagnosed with Williams syndrome without known structural cardiopathy in Argentina in childhood, and he had been living in Spain for a year. On physical examination there were multiple regional axillary hard lymph nodes in the right axilla, greater than 1 cm of diameter, freely movable in the subcutaneous space. No more lymphadenopathies were found.

An axillar ultrasound was performed, demonstrating various hypoechoic lymphadenopathies, greater than 1 cm of diameter (the largest was 3.2 cm), with loss of vascular hilum, and moderate vascularization with Doppler technique. Computed Tomography showed the same findings. Hemogram, kidney and liver function tests, serum proteins, immunoglobulins and antinuclear antibodies were normal. Testing for Human Immunodeficiency Virus, C Hepatitis Virus, B Hepatitis Virus, Epstein Barr Virus, *Cytomegalovirus*, Syphilis and Tuberculosis was negative. A fine-needle biopsy was made, with no findings in cytology and cultures. Therefore, an ultrasound-guided thick needle biopsy was made, with histological findings of vascular and histiocytic proliferation, suggestive of vascular transformation of lymph node sinuses.

Discussion

We present a case of vascular transformation of lymph node sinuses in a patient with previous diagnosis of Williams syndrome in childhood. VTS is an infrequent benign alteration of the lymph nodes, that usually presents as swelling, and its diagnosis is histological, after lymphadenopathy biopsy. Despite its low incidence, it is important to know about its characteristics, and to take it into consideration within the differential diagnosis of enlarged lymph nodes.

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We didn't find any other case reported of VTS associated with Williams syndrome in the literature. VTS is considered to occur secondary to obstruction of the lymph node venous or efferent vessels, but complete physiopathology is not fully understood. There have been findings demonstrative of intussusceptive lymphangiogenesis, and the participation of lymphatic channels (Podoplanin and CD34), myofibroblasts, collagen and lymphodinamic conditions have been postulated [1]. Further, the presence of elastin has been demonstrated in the reticular network of lymph node cortex [4]. On the other hand, elastin is located in the lymph node capsule-associated conduits, and is also involved in fluid and antigen distribution in the skin and lymph nodes [5]. Therefore, the concurrency of Williams syndrome and VTS raises the hypothesis that elastin deficiency may play a role int he pathophysiology of VTS, further research to confirm this hypothesis is warranted.

In conclusion, we present a rare association of axillar lymph node enlargement due to VTS in a patient with Williams syndrome which, to the best of our knowledge, has not previously been reported. Elastin deficiency may play a role in this association.

References

1. Díaz-Flores L, Gutiérrez R, García MP, González-Gómez M, Carrasco JL, Álvarez-Argüelles H. Intussusceptive lymphangiogenesis in vascular transformation of lymph node sinuses. Acta Histochem. 2019;121(4):392-9.

2. Ghosh P, Saha K, Ghosh AK. Vascular transformation of bilateral cervical lymph node sinuses: A rare entity masquerading as tumor recurrence. J Maxillofac Oral Surg. 2015;14(S1):397-400.

3. Pober BR. Medical progress: Williams-Beuren syndrome. N Engl J Med. 2010;362:239-52.

4. Kaldjian EP, Elizabeth Gretz J, Anderson AO, Shi Y, Shaw S. Spatial and molecular organization of lymph node T cell cortex: A labyrinthine cavity bounded by an epithelium-like monolayer of fibroblastic reticular cells anchored to basement membrane-like extracellular matrix. Int Immunol. 2001;13(10):1243-53.

5. Lin Y, Louie D, Ganguly A, Wu D, Huang P, Liao S. Elastin shapes small molecule distribution in lymph node conduits. J Immunol. 2018;200(9):3142-50.