Soft Tissue Rosai-Dorfman Disease
Case report

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Abstract: Rosai-Dorfman disease (RDD) is a rare benign proliferative histiocytic disorder characterised by massive lymphadenopathy. While extranodal involvement can occur in generalised RDD, isolated soft tissue RDD (STRDD) is extremely rare. We report a 17-year-old male patient who presented to the maxillofacial outpatient department of the Sultan Qaboos Hospital, Salalah, Oman, in 2015 with a painless cheek mass which had been slowly growing over the previous two months. Routine histopathological examinations and immunohistochemistry confirmed a diagnosis of STRDD. Currently, surgical excision is considered to be the most effective curative treatment for STRDD, as the outcomes of other treatment modalities are still unknown. Despite its rarity, STRDD should be considered in the differential diagnosis of histiocytic soft tissue lesions.

Keywords: Histiocytosis; Rosai-Dorfman Disease; Emperipolesis; S100 Proteins; Case Report; Oman.

A 17-year-old male patient presented to the maxillofacial outpatient department of the Sultan Qaboos Hospital, Salalah, Oman, in 2015 with a firm painless nodular mass in his right cheek which had been progressively increasing in size over the previous two months. In addition, he had mild proptosis of the right eye. There were no significant findings in the patient’s medical history or laboratory investigations. On clinical examination, the cheek mass measured 4 cm in diameter and was not associated with any lymph node enlargement. The differential diagnosis was of either a neoplastic or inflammatory soft tissue lesion. An excisional biopsy was performed.

The excised mass was formed of two separate pieces of tissue with a firm whitish cut surface. The larger piece measured 1.7 x 0.8 x 0.3 cm and the smaller one measured 1.5 x 0.7 x 0.4 cm. A microscopic examination revealed fibrocollagenous and adipose...
lesion was continuous with another mass measuring 3.2 x 3.4 cm in the right infratemporal fossa, with obliteration of the fat planes. The optic nerve and extraocular muscles were in contact with the mass, but the globe was still intact. Clinically, this lesion was considered to be of the same disease as the cheek lesion and no specific treatment or biopsy was attempted. After two years of follow-up, there was no evidence of disease progression in either lesion.

Discussion

While RDD has been reported worldwide, it is more common among patients of African descent. Up to 80% of cases occur among individuals in the first two decades of life, although other age groups can also be affected; in addition, RDD tends to occur more frequently among males. In the largest known study of RDD, 43% of 423 patients had associated extranodal disease along with lymphadenopathy; in comparison, only 3% of patients had no detectable lymphadenopathy. The skin and the paranasal sinuses are the most common sites for extranodal involvement.

In an extensive review of the literature, Komaragiri et al. identified only 36 STRDD cases reported between 1969 and 2012. The current report adds an additional case to the existing literature concerning this rare entity. Two large case series of STRDD have been reported from the USA; the first described 17 cases of STRDD, of which four presented with lymphadenopathy, whereas the second found that only one of 18 patients with STRDD had associated lymph node involvement from a single node. In both series, STRDD was more common in the trunk and proximal extremities and tended to grow rapidly, although the actual size of the lesion varied. The age of the affected patients also ranged widely and there was a slight female predominance. Multifocal STRDD is extremely uncommon but recurs more frequently than a solitary lesion. The rate of recurrence of STRDD after excision has been reported to range from 21.4–54%.

Immunohistochemistry is key to differentiating soft tissue lesions in order to determine the correct diagnosis. Morphologically, STRDD is similar to nodal RDD in exhibiting the diffuse subcutaneous infiltration of large polygonal histiocytes, which in turn demonstrate emperipolesis predominantly of lymphocytes, although sometimes plasma cells, erythrocytes and neutrophils are also affected. Infiltration of lymphocytes, plasma cells and neutrophils may also occur, along with the histiocytes. However, mitosis and necrosis are rare. While some researchers have...
reported frequent spindling of the histiocytes, others have claimed this is uncommon.\textsuperscript{5,11} In cases with histiocyte spindling, STRDD may be confused with a sarcoma.\textsuperscript{10} In terms of immunohistochemistry, RDD histiocytes are consistently positive for S100 proteins and CD68 and negative for CD1a.\textsuperscript{13} The microscopic appearance of an inflammatory pseudotumour can resemble STRDD; however, the histiocytes of inflammatory pseudotumours are negative for S100. These lesions may reflect a histological continuum.\textsuperscript{14} Another differential diagnosis of STRDD is Langerhans cell histiocytosis, in which the histiocytes are positive for both S100 proteins and CD1a.\textsuperscript{15}

The histogenesis of RDD is still unclear, but may be the result of an aberrant immunoresponse.\textsuperscript{7} The disease has also been associated with lymphoproliferative disorders.\textsuperscript{2} In cases of Hodgkin’s lymphoma, certain cytokines have been found to alter the morphology of histiocytes to that seen in RDD.\textsuperscript{16} A relationship between polyomavirus infections and intra-abdominal STRDD has also been postulated.\textsuperscript{11} Due to its rarity, there is as yet no definitive treatment strategy for patients with STRDD; however, surgical excision has been proven effective in preventing disease recurrence.\textsuperscript{10} Further studies are required to establish evidence-based treatment strategies for this disease and to increase our understanding of its outcomes and natural history.

Conclusion

STRDD is a very rare benign proliferative histiocytic disorder. Morphologically, it may mimic an inflammatory pseudotumour, sarcoma or Langerhans cell histiocytosis. As such, it is important that STRDD be considered in the differential diagnosis of histiocytic soft tissue lesions.

References


