ORIGINAL RESEARCH PAPER

Pathology

A RARE CASE PRESENTATION OF ROSAI-DORFMAN-DESTOMBES DISEASE (RDD)

KEY WORDS: Rosai-Dorfman-Destombes disease (RDD), Lymph node, Histiocyte

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ABSTRACT

Introduction: Rosai-Dorfman-Destombes disease (RDD) is a reactive clonal proliferation of histiocytes with abnormal function leading systemic manifestation **Case report:** A 39 year old male presented with generalized lymphadenopathy. The lymph node are firm in consistency, painless and mobile. Systemic examination is unremarkable. FNAC of these lymph nodes shows polymorphous lymphoid cell population admixed with plenty of macrophages engulfed RBCs, Lymphocytes i.e Emperipolesis. The diagnosis of Reactive lymphadenitis with emperipolesis was given. The lymph node biopsy showed dilated sinuses filled with histiocytes and plenty of macrophages engulfed RBCs, Lymphocytes i.e Emperipolesis. The final diagnosis of histopathology was given as sinus histiocytosis with Emperipolesis. Correlating with clinical findings, The final diagnosis was made as ROSAI DORFMANN DISEASE. **Discussion:** Rosai-Dorfman-Destombes disease (RDD) is a rare histiocytic disease also known as 'sinus histiocytosis with massive lymphadenopathy'. The aetiopathogenesis of RDD is not properly understood. It has been earlier thought to be reactive histiocytic disorder that lacks clonality . **Conclusion:** RDD is one of rare differential diagnosis for lymphoma presenting as generalized lymphadenopathy.

INTRODUCTION:

Rosai-Dorfman-Destombes disease (RDD) is a reactive clonal proliferation of histiocytes with abnormal function leading systemic manifestation. It is a very rare condition with varied presentation. Hereby presenting a case report below.

Case Report:

A 39 year old male presented with multiple swellings below jaw bone on the left side, in front of the left ear, in left side of lower part of the neck and axilla since 3 years. 32-year-old male presented with multiple swellings in upper part of the left side of the neck below jaw bone for 3 years. These swellings developed slowly, initially small in size has gradually progressed to the current size of each measuring about 3cm x 2cm. It is not associated with pain, skin changes, Fever or discharge. Patient had a significant past history of similar swelling in his inguinal region on both sides when he was of age 8-9 year old for which he had undergone excision. On examination, Patient is moderately built and nourished and is well oriented to time, place and person. The swellings appear to be of lymph node which are firm in consistency, painless and mobile.

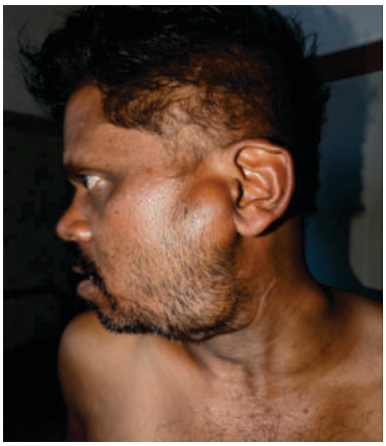

Fig 01: Swellings over left side of face and neck

Systemic examination is unremarkable. On investigation, Complete blood count was normal. Peripheral smear study shows normal blood picture. No blasts were seen. USG of Neck shows hypoechoic lesion of size 2.6 cm – in left parotid gland with multiple cervical lymph nodes, multiple hypoechoic lesion in left submandibular salivary gland. Right parotid and submandibular gland appears normal. FNAC of these lymph nodes shows polymorphous lymphoid cell population admixed with plenty of macrophages engulfed RBCs, Lymphocytes i.e Emperipolesis. The diagnosis of Reactive lymphadenitis with emperipolesis was given.

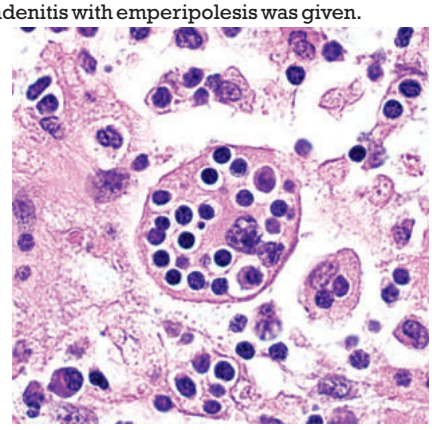

Fig 02: Section studied from lymph node shows emperipolesis(H&E, 1000x)

The lymph node biopsy showed dilated sinuses filled with histiocytes and plenty of macrophages engulfed RBCs, Lymphocytes i.e Emperipolesis. The final diagnosis of histopathology was given as sinus histiocytosis with Emperipolesis. Correlating with clinical findings, The final diagnosis was made as ROSAI DORFMANN DISEASE.

DISCUSSION:

Rosai-Dorfman-Destombes disease (RDD) is a rare histiocytic disease also known as 'sinus histiocytosis with massive lymphadenopathy'. The aetiopathogenesis of RDD is not properly understood. It has been earlier thought to be reactive histiocytic disorder that lacks clonality .However recent researches have shows of clonal expression in few studies. Kinase mutations have been recently seen in ARAF, MAP2K1, NRAS and KRAS. In addition, alterations in genes involved in intracellular trafficking (SNX24), transcriptional regulation (CIC, INTS2, SFR1, BRD4, PHOX2B), cell cycle regulation (PDS5A, MUC4), DNA mismatch repair (ERCC2, LATS2, BRCA1, ATM) and the ubiquitin proteasome pathway (USP35) are also demonstrated.[1]

RDD is classified into sporadic, Familial, and cutaneous forms. The sporadic have nodal, extranodal, Lymphoma associated and Autoimmune associated form. Familial RDD is

characterised by hyperpigmentation, hypertrichosis, hepatosplenomegaly, hearing loss, heart anomalies, hypogonadism, short stature and hyperglycaemia. Cutaneous RDD shows distinct epidemiological and clinical features [2].

The cell of significance is believed to be activated macrophages. These macrophages are activated by the viral agents and modified its function in an abnormal form[3].

Classic RDD presents with massive bilateral painless cervical lymphadenopathy with associated fever, loss of weight and night sweats. It affects most commonly children and young adults with slight male dominance. Lymph nodes affected more commonly are Inguinal, retroperitoneal and mediastinal lymph nodes. Common extranodal sites of involvement include the skin, nasal cavity, bone ,orbital tissue and central nervous system. Bone involvement is seen as lytic lesions. Central nervous system involvement may mimic meningioma. The laboratory findings shows elevated erythrocyte sedimentation rate, leucocytosis, hypergammaglobulinaemia and autoimmune haemolytic anaemia [4].

The classical histological finding in RDD is an enlarged node with a low power appearance of extensive sinusoidal expansion. The cortex comprises numerous activated B cells and mature plasma cells with few follicles which, together with the pale histiocytes, give the appearance of alternating dark and light zones. The sinusoids contain numerous large histiocytic cells showing emperipolesis- intact haematolymphoid cells within a vacuole or floating freely in the cytoplasm of the histiocytes . The histiocytes are S100, CD68 and CD163 positive and are CD1a and langerin (CD207) negative[5,6].

CONCLUSION:

The knowledge regarding a rare histiocyte dysfunction disorder i.e Rosai-Dorfman-Destombes disease (RDD) should be accumulated in vast case series to know the exact etiology and presentation.

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