ROSAI-DORFMAN DISEASE: REPORT OF SIX CASES AND REVIEW OF LITERATURE

MALADIE DE ROSAI-DORFMAN : À PROPOS DE 6 CAS ET REVUE DE LA LITTÉRATURE

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ABSTRACT

Introduction: Rosai Dorfman disease is a benign non-Langerhansian histiocytosis. Its etiology remains unknown. It consists of the presence of histiocytes within lymph nodes, but other extra-nodal localizations such as nasosinusal or orbit may be noted. It frequently affects young adults and adolescents. Diagnosis is based on anatomopathological evidence. The aim of this study was to specify the clinical and therapeutic features of this disease.
Methods: We conducted a retrospective descriptive study of 06 patients diagnosed with Rosai Dorfman disease in the ear, nose and throat (ENT) department of Habib Thameur Hospital Tunisia over a 07-years period from 2017-2023.
Results: Six patients were diagnosed with Rosai Dorfman disease in our department. The mean age was 18.7, with extremes ranging from 9 months to 66 years and a sex ratio (M/F) of 1. The mean time from symptom onset to diagnosis varies from 2 months to 12 months. Two patients had single lymph node involvement, 2 had isolated nasosinusal involvement, one had nasosinusal involvement associated with pulmonary involvement, and one had lymph node involvement associated with laryngeal, parotid, cutaneous and mammary involvement. The diagnosis of certainty was made on definitive histopathological examination with immunohistochemical study. Two patients required corticosteroid therapy at a dose of 1mg/kg/d combined with an immunosuppressive drug such as methotrexate (15mg/week).
Conclusion: The diagnosis of Rosai Dorfman disease relies on histopathological examination Although rare, it is an entity that should not be ignored to ensure better therapeutic management.
Key words: Rosai-Dorfman disease, Histiocytosis, Lymphadenopathy nodal, extranodal.

RÉSUMÉ

Introduction : La maladie de Rosai dorfman ou histiocytose sinusale avec lymphadenopathie est une histiocytose bénigne non langerhansienne. Elle demeure d'étiologie inconnue. Elle est caractérisée par la présence d'histiocytes au sein des chaines ganglionnaires mais d'autres localisations extra ganglionnaires telle que nasosinusiennes ou orbitaire peuvent être notées. Elle touche très souvent l'adolescent et l'adulte jeune. Le diagnostic est posé sur des preuves anatomopathologique. Cette étude a pour objectif d'étudier les caractéristiques clinico-histologique de la maladie de rosai dorfman à localisation ORL.
Matériels et méthodes : il s'agit d'une étude rétrospective descriptive colligeant 06 patients diagnostiqué pour la maladie de rosai dorfman au service d'ORL de l'hôpital Habib Thameur sur une période de 07 ans allant de 2017-2023. Nous avons inclus les patients présentant une atteinte ganglionnaire cervicale ou extra ganglionnaire ORL.
Résultats : L'âge moyen de nos patients était de 18,7 ans, avec des extrêmes allant de 9 mois à 66 ans. Le sexe ratio était de 1. Le délai moyen entre l'apparition des symptômes et le diagnostic variait de 2 à 12 mois. Deux patients présentaient une atteinte lymphatique unique, deux patients une atteinte nasosinusale isolée, un patient une atteinte nasosinusale associée à une atteinte pulmonaire et un patient une atteinte lymphatique associée à une atteinte laryngée, parotidienne, cutanée et mammaire. Le diagnostic définitif a été posé après un examen histopathologique avec étude immunohistochimique. Deux patients ont nécessité un traitement par corticostéroïdes à une dose de 1mg/kg/j associé à un immunosuppresseur par le méthotrexate (15 mg/semaine).
Conclusion : le diagnostic d'une maladie de rosai dorfman repose sur l'examen histopathologique avec étude immunohistochimique. bien que rare c'est une entité à ne pas méconnaitre afin d'assurer une meilleure prise en charge thérapeutique.
Mots clés : maladie Rosai-Dorfman, Histiocytose, Lymphadenopathie, extra ganglionnaire.

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INTRODUCTION:

A benign systemic histio proliferative disease, Rosai-Dorfman disease (RDD) is also referred to as sinus histiocytosis with massive lymphadenopathy [1]. Clinicians and pathologists need to be aware of it because it is one of the differentials of cervical lymphadenopathy. Although RDD is an idiopathic condition, it has often been found to emerge after infection. Several papers have proposed that Epstein-Barr virus (EBV), human herpes 6, parvovirus B19 (HHV-B19), and polyomavirus could be the cause of this condition [2] . Though extranodal locations of the ENT have also been documented, RDD always includes lymph nodes. Six RDD cases including ENT involvement that were dealt with in our ENT department were examined in this study.

METHODS:

We conducted a retrospective descriptive study of 06 patients diagnosed with Rosai Dorfman disease in the ENT department of Habib Thameur Hospital Tunisia over a 07-year period from 2017-2023. We included patients with nodal or extranodal ENT involvement.

From clinical records, general data was retrieved, including gender, the outset of the disease, symptoms, imaging findings, pathological information, and given medical treatment.

RESULTS:

Six patients were diagnosed with Rosai Dorfman disease in our department. The mean age was 18.7, with extremes ranging from 9 months to 66 years. The sex ratio was 1. The mean time from symptom onset to diagnosis varies from 2 months to 12 months.

The initial reason for consultation was an isolated laterocervical tumefaction of chronic evolution in 2 patients, a laterocervical tumefaction of chronic evolution associated with exertional dyspnea in one patient, headache associated with hypoacusis and ringing in one patient, nasal obstruction in one patient and multifocal orbital and nasosinusal cysts. In addition to palpable cervical adenopathy in 3 patients, the clinical examination revealed the presence of formation in the posterior wall of the glottis, as assessed by indirect laryngoscopy in the 3rd patient, a bulging of the left posterolateral wall of the nasopharynx bleeding on contact in the patient who presented with headache, and nasal fossa formation in the last two patients.

The characteristics of the studied patients are shown in table 1.

Table 1: Initial characteristics of the studied patients

	1	2	3	4	5	6
Age	6 years	3 years	25 years	66 years	9 months	12 years
Sex	F	H	F	H	H	F
Length of symptom onset (months)	12	2	2	12	9	12
Head and Neck Site	Cervical lymph node	Cervical lymph node	Cervical lymph node + Subglottis	Nasopharynx + Paranasal sinuses + lung	Paranasal sinuses	Paranasal sinuses

Radiological investigations were undertaken in all cases. Neck ultrasound was performed in the 2 cases who had single lymph node involvement. Sinus CT scan and sinus MRI were carried out in 2 cases who had nasosinusal involvement (figure 1+2). Cervico-thoraco-abdomino-pelvic CT was performed in both patients with multiple sites (figure 3).

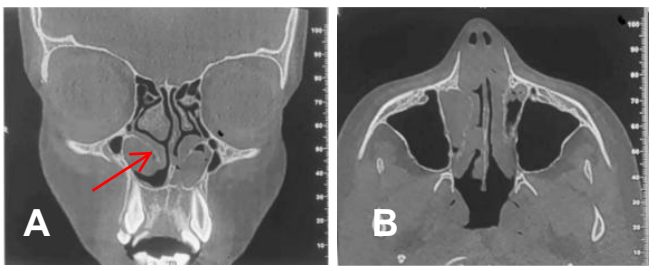

Figure 1: (A) + (B) CT coronal and axial section showing polypoid formations in the nasal cavities of the lower meats, which seem to emerge from the lacrymonasal duct .

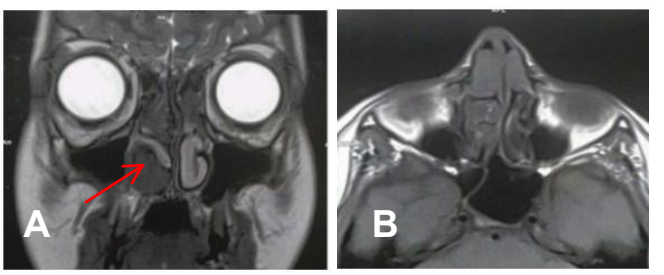

Figure 2: (A) + (B) MRI coronal + axial section showing suspicious nasal cavity process (decreased ADC)

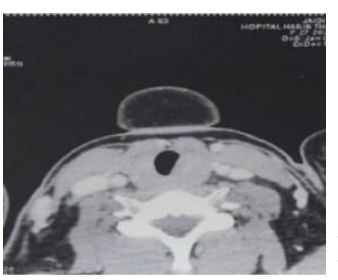

Figure 3: CT axial section showing subglottic laryngeal tissue nodule lateralized to the left

Fine needle aspiration was performed in two patients, concluding that lymphoid cells were present which were difficult to type. Surgical management consisted of adenectomy in the first two cases, adenectomy with direct laryngoscopy in the 3rd case, and later tracheotomy when the patient presented with high dyspnoea on awakening. One patient had cavum biopsy on several occasions, with intraorbital biopsy. The last two patients underwent endonasal exeresis of nasal fossa formations (figure 4).

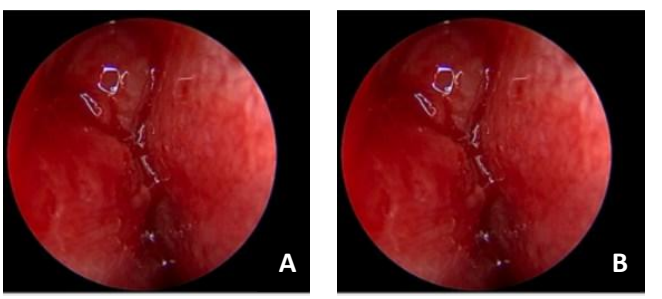

Figure 4: -A: intraoperative endoscopic appearance of a nasal fossa polyploid mass in Rosai Dorfman disease -B: exeresis of nasal mass

Extemporaneous examination was inconclusive in all cases. The diagnosis of certainty was made on definitive histopathological examination with immunohistochemical study. A locoregional and distant extension study was carried out, concluding that 2 patients had single lymph node involvement, 2 had isolated nasosinusal involvement, one had nasosinusal involvement associated with pulmonary involvement, and one had lymph node involvement associated with laryngeal, parotid, cutaneous and mammary involvement.

The two patients with more than one affected site required corticosteroid therapy (1mg/kg/d prednisone) associated with an immunosuppressant such as methotrexate 15mg/week.

The average follow-up was 5,3 years ranging from one year to 7 years.

The evolution was good in 5 cases, one patient with nasopharyngeal and pulmonary involvement died.

DISCUSSION:

Rosai Dorfman disease (RDD) was first described by the French pathologist Paul Destombes in 1965 [3]. It is a rare nonmalignant type of histiocytosis of unknown cause. It occurs more frequently within young adults [4]. In our study 5 patients were under 25 years of age when first diagnosed. Although in our study the sex ratio was 1, a male preponderance has been reported in the literature [5]. Bilateral painless cervical lymphadenopathy is the most frequent symptom, occurring in 90% of patients [6]. In our group of patients, 3 of the 6 had palpable cervical adenopathy which represents 50% of cases. Adenopathy can be axillary(38%), inguinal (44%), hilar and mediastinal (40%), and even retroperitoneal which are rare [7,8]. The lymphadenopathy may be associated with fever and night sweats [5]. Extranodal involvement, with or without nodal involvement, is noted in 40% of cases [9,10]. The involvement of the nasal cavity and paranasal sinuses reported in literature is 11% of RDD cases [9] . The most frequent symptoms are nasal obstruction, epistaxis with the presence on examination of lesions in the nasal cavity, paranasal cavity or even the nasopharynx. Even though these symptoms are nonspecific, they underline how important it is to take RDD into consideration when making a differential diagnosis of nasal masses, especially when recurrent or atypical signs are present [11]. In addition, clinical manifestations can be easily misdiagnosed as nasal tumors [12]. RDD with laryngeal involvement is rare less than 30 such cases have been reported [13]. The main clinical signs include foreign body sensation, hoarse voice, dyspnea, and cough [14]. Like sinus involvement, symptoms in laryngeal involvement are non-specific. However, it is a life threatening location because the lesion may lead to respiratory tract obstruction [15,16].

The radiological manifestations of RDD vary according to organ involvement, and its imaging features are often confused with other diseases [17]. Imaging contributes to the diagnosis by identifying the differential diagnoses. Fine needle aspirate (FNA) conclude often to non-specific inflammatory entity [18]. The limitations of cytology are due to the fibrosis found in RDD [19]. The diagnosis is made by anatomopathological examination of biopsy or surgical resection tissues [20]. Histological examination reveals large cells with a broad eosinophilic or sometimes xanthomized cytoplasm ("Destombes cells") and large nuclei. The almost pathognomonic image is the presence of numerous intact lymphocytes in the cytoplasm of the histiocytes [21]. RDD is characterized by the following phenotype: S100+,CD1a-, CD68++, Fascin++, CD163++ [8].

There are not any specific treatment modalities. It has been reported that 20% of patients showed spontaneous resolution [22]. When anatomically feasible, complete excision is the treatment of choice [5]. Although results have differed, corticosteroids are generally beneficial in lowering nodal size and symptoms. Prednisone is the most suitable corticosteroid [9]. Methotrexate and other chemotherapy are often saved for cases that are refractory or relapsed, while they can occasionally be employed as the first line of treatment for diseases that are disseminated or life-threatening [9]. Radiotherapy and immunomodulators have been described in the literature. The limited group of patients makes it unsuitable for any therapeutic approach.

The prognosis of lymph node RDD is favorable in most cases. However, there are no large case series to assess the prognosis of patients with RDD. In extranodal forms of RDD, prognosis also correlates with the number of organ systems concerned [23].

CONCLUSION:

A benign, pseudolymphomatous entity, Rosai-Dorfman disease may appear in a variety of manifestations throughout the head and neck. Cervical lymph node involvement is the most distinctive finding reported in the literature; however, extranodal manifestations have been widely described. The diagnosis is based on anatomopathological examination. The prognosis is generally favorable. There is no consensus regarding treatment. As the etiology remains obscure, further work is needed.

Data availability statement:

The data that supports the findings of this study are available upon request.

Ethical Statement:

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request. ethical approval was given by the ethical committee of Habib Thameur Hospital – T0849

Conflict of interest: The authors declare having no conflicts of interest regarding this manuscript.

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