A Rare Case of Rosai-Dorfman Disease of the External Acoustic Meatus Mimicking Otitis Externa

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ABSTRACT

Rosai-Dorfman Disease is a rare condition that can manifest in almost any part of the body. It commonly presents as intranodal disease, with less common extra-nodal involvement tending to occur in the head and neck region. We present a case of Rosai-Dorfman Disease involving the external acoustic meatus in a patient and will detail its imaging and clinical features. We will also discuss its imaging characteristics and compare them to other common conditions such as otitis externa and squamous cell carcinoma. In doing so, we hope to provide more information about this rare disease.

CASE REPORT

CASE REPORT

A 28-year-old male patient presented with reduced hearing in the right ear. He had no significant past medical history and denied any associated pain or discharge. On examination, there was swelling of the external acoustic meatus (EAM). The diagnostic impression was otitis externa and the patient was discharged with a trial of antibiotic eardrops. Symptoms did not improve and the patient returned 3 weeks later. This prompted a referral to an otolaryngologist.

Further examination revealed a closed up and edematous EAM (Figure 1A). Otoscopy revealed a dull tympanic membrane with a pale whitish appearance of the EAM when compared to the unaffected left ear (Figure 1B,1C). This was unusual for otitis externa, however the patient was placed on a trial of Combiderm (clotrimazole 1%, beclomethasone dipropionate 0.025%, and gentamicin sulfate 0.1%) which helped with his symptoms and he was subsequently discharged from follow-up.

Three months later, the patient presented again with a recurrence of symptoms. There was swelling of the tragus with a “cystic” consistency. Incision and drainage was attempted, however, no pus or discharge was observed. A fine-needle aspiration (FNA) was performed to collect samples for further analysis. Inflammatory blood markers (including white blood cell count and C-reactive protein) were found to be normal.

Preliminary FNA results showed atypia of undetermined significance which prompted further evaluation with an excisional biopsy due to concerns of an underlying malignancy. However, prior to the excision biopsy, MRI of the EAM was performed to further characterize and assess the extent of the disease.

MRI showed a well-encapsulated homogenous circumferential soft tissue thickening of the right EAM which appeared T1-weighted isointense (Figure 2A), T2-weighted fat saturated sio-hyperintense (Figure 2B) with homogeneous contrast enhancement (Figure 2C,2D) and minimal restricted diffusion (Figure 2E,2F). A small right mastoid effusion and prominent right cervical lymph nodes were also seen.

There was minimal surrounding fat stranding seen on imaging. Clinically, apart from swelling, there was no pain, erythema or discharge that is commonly seen with infection, in fact, the EAM had a pale appearance with squamous debris. This made otitis externa less likely. Several mildly prominent

cervical lymph nodes and a small right mastoid effusion were also seen on the MRI, however this remained non-specific.

Neoplastic processes such as squamous cell carcinoma, adenoid cystic carcinoma and lymphoma were also considered. However, these are usually aggressive diseases that demonstrate trans-spatial and perineural spread [1]. Lymphoma would also demonstrate significant restricted diffusion. A cholesteatoma is less likely given that the location within the EAM is rare and it does not demonstrate significant restricted diffusion which is typically seen on imaging [1]. Vascular lesions were also considered, however, no significant vessels or flow voids were seen in the lesion. Given these findings, an underlying infiltrative disease remained a possibility.

Intraoperatively, a fleshy whitish-yellow lesion seen around the external acoustic canal and tragus with a well-formed capsule, this extends to the tympanic ring. An adjacent lymph node was also excised.

Histology shows large histiocytic cells which are positive for S-100 protein and CD 163 but negative for CD1a. There is also presence of fibrosis. Findings are consistent with Rosai-Dorfman Disease (RDD).

DISCUSSION

Etiology and demographics

RDD is a rare non-Langerhan cell histiocytic disease characterized by accumulation of activated histiocytes within the affected tissue. The prevalence of this disease is 1:200 000 and estimated 100 new cases per year in the United States [2]. It is more frequently seen in children and young adults (mean age, 20.6 years) as well as males of African descent, but cutaneous forms more commonly present in Asians [2]. It can occur in isolation or in association with autoimmune or malignant diseases such as IgG4 disease, systemic lupus erythematous, idiopathic juvenile arthritis, autoimmune haemolytic anaemia and HIV [2].

It usually presents as intranodal disease. Extranodal disease manifestation is rarely seen without nodal disease. The most frequent extra-nodal sites were found to be in the head and neck region, for example - skin and soft tissue of the head and neck as well as other parts of the body (16%); nasal cavity and paranasal sinuses (16%); eye, orbit, and ocular adnexa (11%) [3]. To the best of our knowledge, there is no publication that describes similar EAM involvement as seen in our case. A review of RDD cases performed by Vaidya, Mahajan and Rane did not reveal any case involving the EAM [4]. There was only a case of cutaneous involvement of the pinnae as described by Raslan, Schellingerhout, Fuller, Ketonen [5].

Histologically, RDD is distinguished not only by the proliferation of histiocytes, but also by their ability to engulf intact lymphocytes and other immune cells, a feature known as emperipolesis [2]. The diagnosis is confirmed through the detection of specific markers on histiocytes, such as S100, CD68, and CD163, while CD1a staining and BRAF V600E mutations are typically absent [2]. These histological and immunohistochemical characteristics help differentiate RDD from Langerhans cell histiocytosis and Erdheim-Chester Disease, which lack emperipolesis and instead exhibit Langerhans cells and foamy histiocytes [3]. The pathogenesis of this disease is not currentl known. Human herpesvirus 6, parvovirus B19 and Epstein-Barr virus, have been investigated as potential inciting agents, however, studies have failed to determine a causative link [2].

Clinical and imaging findings

It is very difficult to distinguish this disease from other common pathologies of the EAM based on imaging alone as they share many similar characteristics. In this case, the clinical presentation is that of a “cold” circumferential lesion of the external acoustic meatus that did not respond to antibiotics, but to a combination cream containing steroid additives.

The imaging features we observed in this case were similar to that observed in the current literature [4, 5]. However, these imaging features are also commonly seen in otitis externa, cholesteatoma and malignancies of the EAM, such as lymphoma, adenoid cystic carcinoma or squamous cell carcinoma (Tables 1,2). Hence, it is difficult to tell purely based on imaging that this was RDD, correlation with clinical presentation and examination findings is crucial. The fact that there was no trans-spatial involvement of the disease makes malignancy less likely, there was also no surrounding inflammatory change such as fat stranding that pointed towards a diagnosis of infection. Another possible differential was relapsing polychondritis, however, this disease is often bilateral and usually presents clinically with painful swelling and erythematous pinnae with sparing of the ear lobes [1].

That being said, a patient with RDD could have superimposed infection. In our patient, swabs of the EAM demonstrated growth of Pseudomonas aeruginosa which could have swayed the diagnosis to otitis externa if one is not discerning. Hence, this emphasizes the importance of correlation with clinical presentation and symptoms.

Once diagnosed, a patient will require further evaluation with PET CT to determine the extent of disease. Our patient underwent a PET CT scan which demonstrated RDD involvement of the right EAM (Figure 3A), nodal involvement in the mediastinum and cutaneous involvement (Figure 3B) along the right chest wall (Figure 3C).

Treatment and prognosis

Sporadic RDD typically has a good prognosis, with about 50% of cases experiencing spontaneous remission [6]. However, up to 10% of patients might die from complications such as infections or amyloidosis [6]. Current guidelines suggest that observation can be performed for uncomplicated lymph node involvement and asymptomatic skin disease [6]. Surgical excision should be considered for localized extranodal disease

or symptomatic cases affecting the airway, cranial, spinal, or sinus areas [6]. Systematic therapy can be used for multifocal, irresectable extranodal disease. Although there is no standardized treatment regimen, options include corticosteroids, sirolimus, radiotherapy, chemotherapy, and immunomodulatory therapy [6]. For severe refractory cases, next-generation sequencing for MAPK mutations is advised, with potential targeted therapy if driver mutations are found [6].

TEACHING POINT

RDD of the EAM is rare, however, it needs to be considered in cases with atypical symptoms. Its imaging findings often mimic common conditions such as otitis externa and malignancy, however, imaging features such as a well-encapsulated mass without trans-spatial involvement and clinically "cold" appearance helps differentiate this from infection or malignancy.

QUESTIONS & ANSWERS

1. Which of the following imaging characteristics is typically observed in Rosai-Dorfman Disease of the external acoustic meatus on MRI?

A) Diffuse, ill-defined mass with significant surrounding fat stranding

B) Well-encapsulated mass with T1-weighted hypointensity, T2-weighted iso-hyperintensity, and minimal diffusion restriction (applies)

C) Hypervascular lesion with internal flow voids

D) Rapidly growing mass with bony erosion and diffusion restriction

E) T1w hyperintense, T2-weighted hypoeintense mass with restricted diffusion.

2. How does Rosai-Dorfman Disease clinically differ from otitis externa or malignancy in the external acoustic meatus?

A) Rosai-Dorfman Disease typically presents with a painful, erythematous mass with discharge, whereas otitis externa presents with a painless, well-encapsulated mass.

B) Rosai-Dorfman Disease often appears as a non-tender, well-encapsulated mass with pale appearance and squamous debris, while otitis externa is characterized by painful swelling, discharge, and tenderness. (applies)

C) Rosai-Dorfman Disease presents with rapid growth and systemic symptoms, whereas malignancies typically present with a non-tender mass with minimal local symptoms.

D) Otitis externa is associated with well-defined, hypervascular lesions with internal flow voids, whereas Rosai-Dorfman Disease shows significant diffusion restriction on MRI.

E) Rosai-Dorfman Disease are slow growing vascular lesions that tend to demonstrate flow voids and significant diffusion restriction.

3. Which of the following best describes the histological hallmark of Rosai-Dorfman Disease?

A) Presence of Langerhans cells and foamy histiocytes

B) Emperipolesis, or histiocyte-mediated phagocytosis of intact lymphocytes (applies)

C) Positive CD1a staining and BRAF V600E mutations

D) Significant foamy histiocytes and absence of emperipolesis

E) Negative S100, CD68, and CD163 staining.

4. Which treatment strategies are recommended for multifocal, irresectable extranodal Rosai-Dorfman Disease in a symptomatic patient?

A) Immediate surgical excision

B) Systemic therapy, including corticosteroids, sirolimus, chemotherapy, and immunomodulatory therapy (applies)

C) Observation without further intervention

D) Localized radiation therapy (applies)

E) Ablation of the lesion

5. What key imaging feature is more consistent with Rosai-Dorfman Disease as compared to otitis externa and malignancies in the external acoustic meatus?

A) Presence of surrounding fat stranding

B) Trans-spatial spread and bony erosion

C) Well-encapsulated, homogenous soft tissue thickening without trans-spatial involvement (applies)

D) Increased vascularity and internal flow voids

E) Significant restricted diffusion

AUTHORS' CONTRIBUTIONS

Calvin Cheong is the primary author responsible for writing this manuscript.

Chee Kwang Kee has contributed in describing the imaging findings and editing this manuscript.

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DISCLOSURES

No financial disclosures or conflicts of interests to declare.

CONSENT

Did the author obtain written informed consent from the patient for submission of this manuscript for publication? YES

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FIGURES

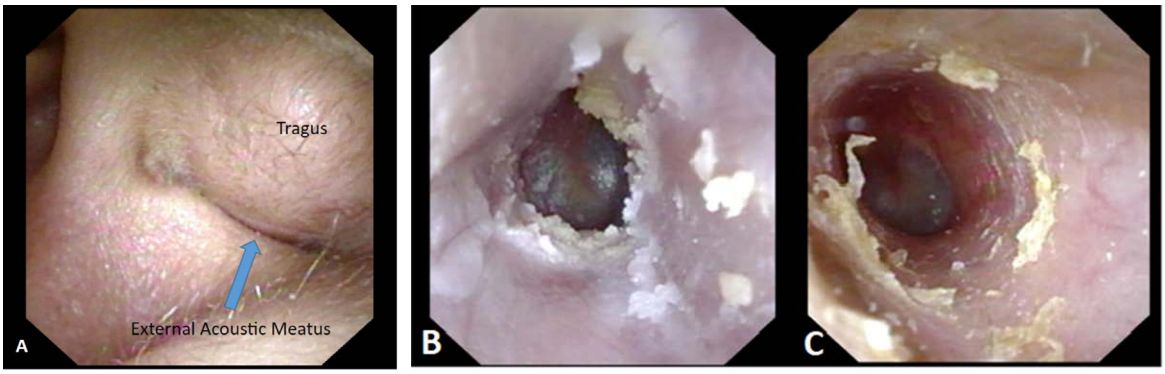

Figure 1: (A) Closed up external acoustic meatus (blue arrow). (B) Otoscopy of the affected right ear demonstrating a pale external acoustic meatus with a dull tympanic membrane surrounded by squamous debris. (C) Otoscopy of the unaffected left ear for comparison.

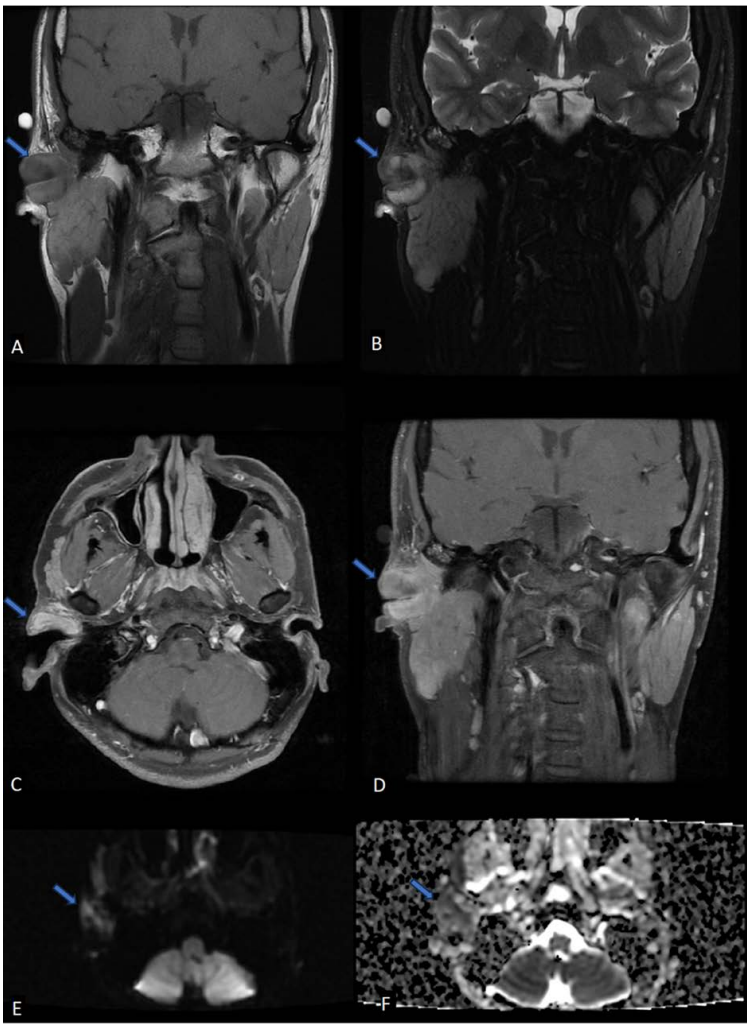

Figure 2: Findings: MRI performed showed circumferential thickening of the right EAM (blue arrow). (A) T1-weighted coronal demonstrating isointense signal; (B) T2-weighted fat saturated coronal demonstrating iso-hyperintense signal; (C) axial post IV-gadolinium contrast T1-weighted fat-saturated shows homogenous enhancement; (D) coronal post IV-gadolinium T1-weighted fat-saturated shows homogenous enhancement; (E) DWI and (F) ADC demonstrating minimal diffusion restriction. *TECHNIQUE:* 3.0T GE Discovery MR 750W; T1w (TE: 12.9, TR: 619.0); T2-fat saturated (TE: 94.1, TR 3482.0); T1w post contrast (TE: 9.9, TR: 833.0); DWI & ADC (TE: 64.9, TR: 3200.0).

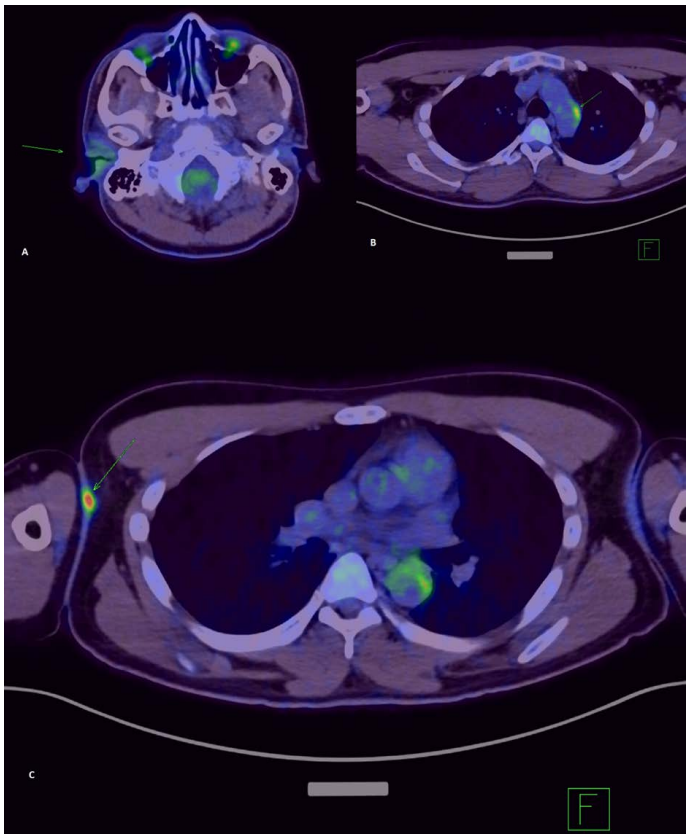

Figure 3: ^{18}F fluorodeoxyglucose PET CT demonstrating avid FDG uptake in the right external acoustic meatus (A), para-aortic node (B) and skin along the right chest wall (C). *TECHNIQUE:* 5.87 mCi of F18 Fluorodeoxyglucose; CT – 100.0kV; Automatic Exposure Control: average 330.0 mA; Slice thickness 3.0mm

SUMMARY TABLE

Table 1: Summary table for RDD of the external acoustic meatus

Etiology	Non-Langerhan cell histiocytic disease characterized by accumulation of activated histiocytes within the affected tissue.
Prevalence	Rare. Prevalence of this disease is 1:200 000 and estimated 100 new cases per year in the United States
Gender Ratio	Slight male predominance (male to female ratio of 1.4)
Age predilection	Children and young adults (mean age, 20.6 years)
Risk factors	No convincing risk factors due to rarity of this disease, but can occur in association with IgG4 disease, SLE, idiopathic juvenile arthritis, autoimmune haemolytic anaemia and HIV
Treatment	Observation for asymptomatic patients. For symptomatic patients, localized disease involving the airway, cranial, spinal, or sinus areas can be excised. Systematic therapy for multifocal, irresectable extranodal disease, using options like corticosteroids, sirolimus, radiotherapy, chemotherapy, and immunomodulatory therapy can be performed although there is no standardized regimen For severe refractory cases, next-generation sequencing for MAPK mutations is advised, with potential targeted therapy if driver mutations are found
Prognosis	Sporadic Rosai-Dorfman disease typically has a good prognosis, with about 50% of cases experiencing spontaneous remission. However, up to 10% of patients might die from complications such as infections or amyloidosis.
Findings on imaging	Findings on MRI include a well-encapsulated homogenous circumferential soft tissue thickening of the right EAM, appearing T1-weighted isointense, T2-weighted fat saturated iso-hyperintense with homogeneous contrast enhancement and minimal restricted diffusion.

DIFFERENTIAL TABLE

Table 2: Comparison of the clinical features and MRI imaging features of the various diseases affecting the EAM

Differential diagnosis	Clinical features	Imaging features on MRI
Rosai Dorfman Disease	Well-encapsulated pale painless mass, no discharge or erythema. Squamous debris in the external acoustic meatus, dull looking tympanic membrane. Enlarged lymph nodes.	Well-encapsulated T2w fat saturated iso-hyperintense. T1w isointense Homogenous contrast enhancement No trans-spatial or perineural spread Bony erosion is rare
Otitis Externa	Erythematous, painful swelling. Possible discharge. Middle ear effusion. Fever. Ipsilateral enlarged tender cervical lymph nodes.	Ill-defined borders Surrounding oedema/fat stranding T2w hyperintense, T1w hypointense Contrast enhancement with diffusion restriction Trans-spatial involvement in severe cases Bony erosion Enlarged lymph nodes Associated features as thrombophlebitis, cerebral abscesses, meningitis, carotid artery pseudoaneurysm
Malignancy such as SCC, adenoid cystic carcinoma or lymphoma	Painful mass with rapid growth. May have overlying skin changes ulceration. Enlarged cervical lymph nodes, usually unilateral	T2w/STIR hyperintense mass T1w hypointense Minimal surrounding oedema or fat stranding Contrast enhancement with diffusion restriction Trans-spatial involvement Bony erosion Enlarged lymph nodes
Relapsing Polychondritis	Usually involves bilateral auricles. Bilateral (or less commonly unilateral) auricular chondritis and polyarthritis Acute painful inflammatory crises are followed by spontaneous remission of variable duration.	Non-specific features of inflammation such as: - Edematous - T2w hyperintense - T1w hypointense - Contrast enhancement Distribution and clinical history are key for diagnosis.
Benign vascular lesions	Increased vascularity of the region with overlying skin discoloration.	Presence of internal flow voids T1w hypointense T2w hyperintense
Cholesteatoma	Slow growing mass in the inner ear, usually at typical locations such as	T2w hyperintense (similar to CSF) T1w hypointense Presence of diffusion restriction

KEYWORDS

Rosai-Dorfman Disease; External acoustic meatus; Ear; Non-Langerhan cell histiocytosis; Head and neck

ABBREVIATIONS

EAM = EXTERNAL ACOUSTIC MEATUS
FNA = FINE NEEDLE ASPIRATION
RDD = ROSAI-DORFMAN DISEASE
DWI = DIFFUSION WEIGHTED IMAGING
ADC = APPARENT DIFFUSION COEFFICIENT