Rosai-Dorfman-Destombes Disease in adolescence with hearing and vision loss: a multidisciplinary involvement

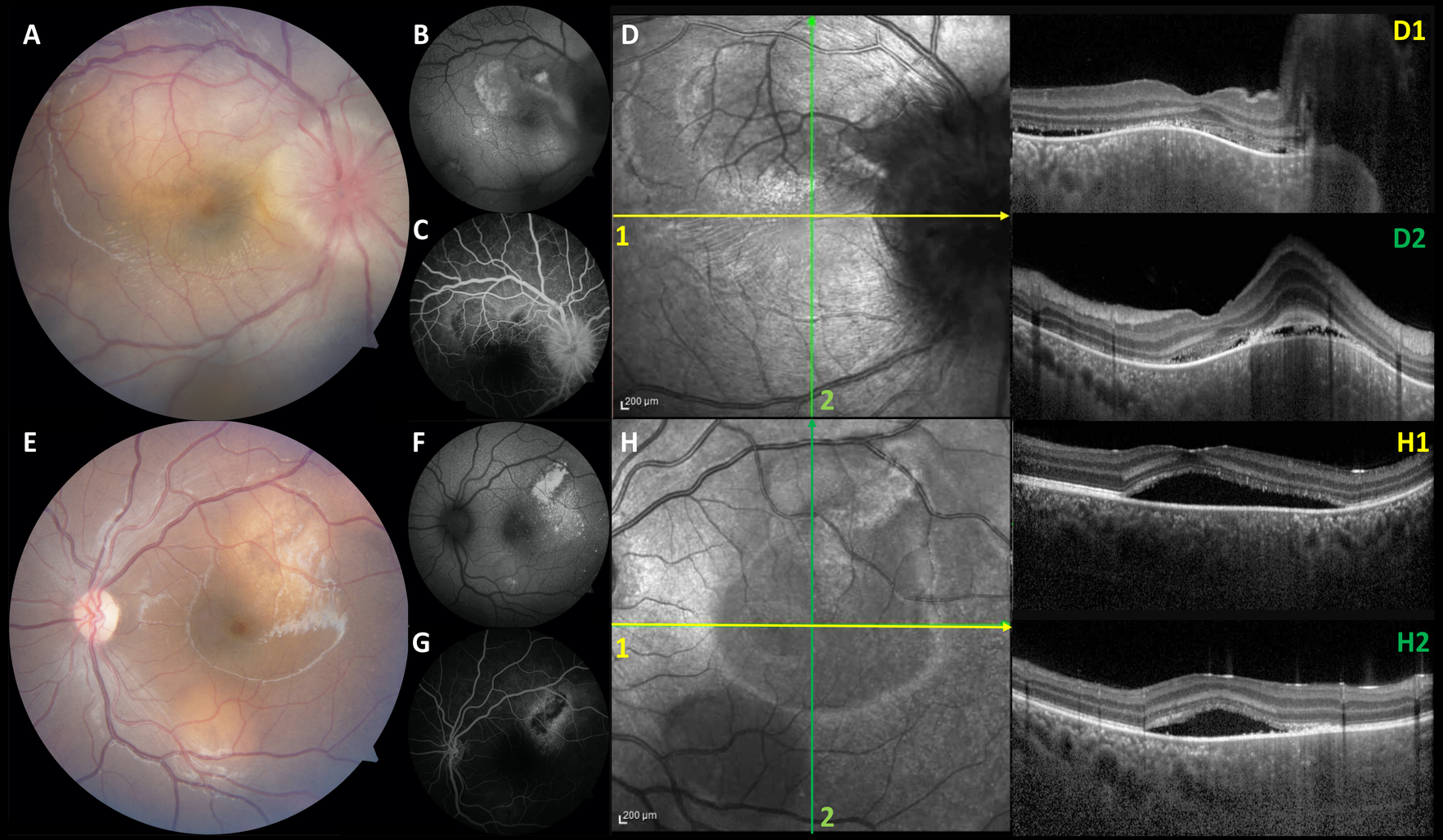
[1]Ophthalmology, São José do Rio Preto medical School (FAMERP), SP, Brazil.
[2]Department of Ophthalmology, University of São Paulo, SP, Brazil.

Gabriela Hamra Pereira, M.D[1],
Marina Gaglianone Teodoro da Silva, M.D[1],
Inara Figueiredo Jacobsen M.D[1],
Maria Nathália Vilela Assis M.D[1],
Antônio Carlos Pondé Rodrigues Jr, M.D[1],
Murilo Wendeborn. Rodrigues Jr., M.D, Ph.D[1],
Eduardo Cunha de Souza, M.D, Ph.D[1,2]

PURPOSE: To report an atypical case presentation of Rosai-Dorfman-Destombes disease, hystiocitosis with predominance of choroidal and neurological involvement. A challenging diagnostic in mimicking other serious diseases like infections (e.g. tuberculosis and sarcoidosis), metastasis, linfoproliferative and autoimune-inflammatory disorder (e.g. VKH).

METHODS: **C**ase presentation of 15-year-old girl complaining of indolent vision loss in OU eyes in the last 3 months. Associated complaims were headache, night sudoresis, hearing loss and tinnitus. BCVA was 20/60 in the OD eye and hands-motion in the OS eye. Ocular anterior segment examination was unremarkable, including IOP. Fundus examination revealed yellow-whitish choroidal lesions temporally, upper and under the fovea in OU eyes, and ON edema in OD (fig. 1AE). Ocular multimodal imaging was performed including near-IR (fig. DH), FA (fig.CG) AF (fig. BF), SD-OCT (FIG. D1-2, OD; H1-2, OS). A multidisciplinary evaluation was request with an extensive systemic work-out including BT and whole-body imaging with MRI and PET-scan to rule out other differential etiologic diagnoses (figs. 2,3).

FIGURE 1: ocular multimodal imaging.



ABREVIATIONS: RDD, Rosai-Dorfman-Destombes; VKH, Vogt-Koyanage-Harada; OU, oculus uterque; BCVA, best corrected vision acuity; OD. Oculus dexter, OS, oculus sinister; IOP, intraocular pressure; ON, optic nerve, IR, near-infrared reflectance; FA, fluorescein angiography; AF, autofluorescence; VF, visual-field; SD-OCT, spectral-domain optical coherence tomography; USS, ultrasound-scan; BT, blood-tests; MRI, magnetic ressonance imaging; PET, positron emission tomography.

FIGURE 2. MRI imaging: (A,F) choroid plexus hipersign. (B,G) pre-bulbar mass. (C) pre-quiasmatic thickning. (D) cerebellopontine angle thickning. (H) internal acoustic canal realce. (I) basal frontal pachymeninge plaque. (E,J) etmoidal and maxilar paranasal sinuses mass.

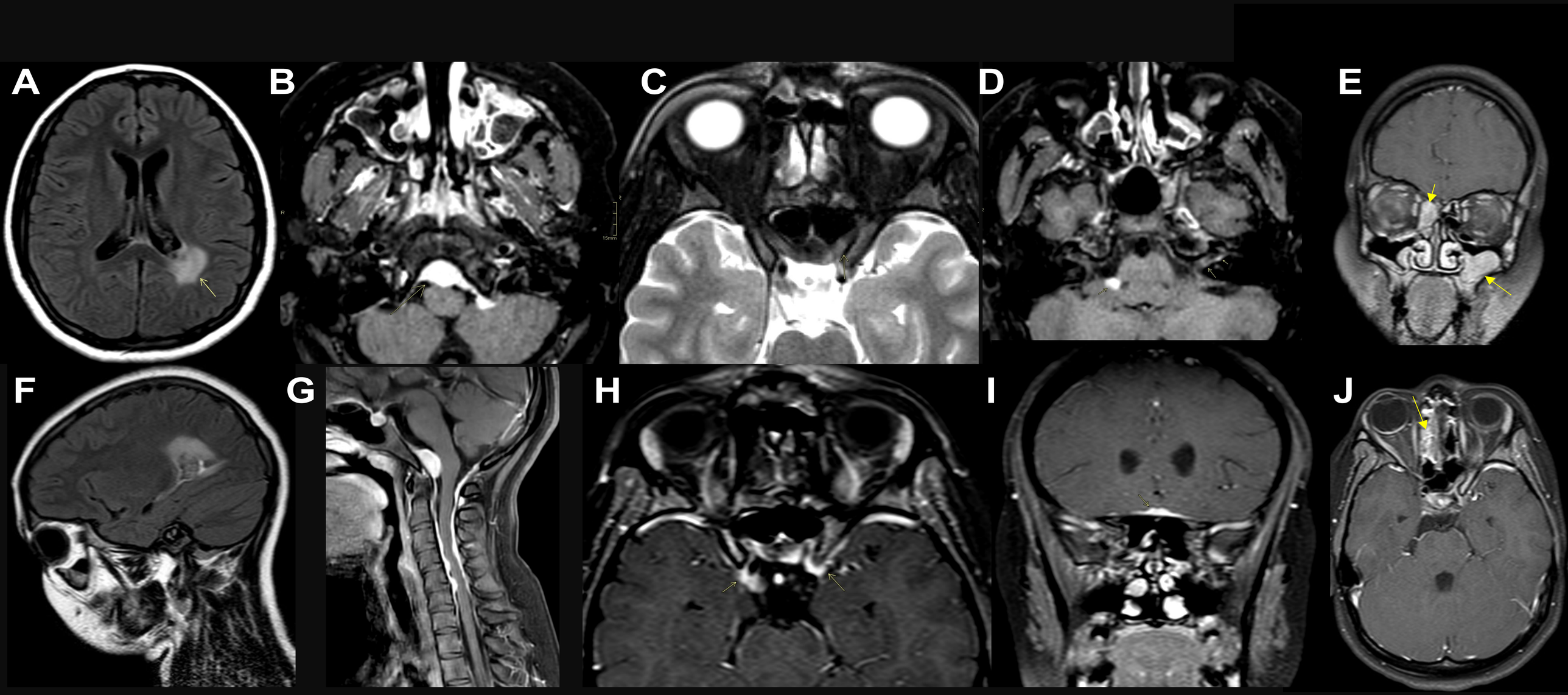


FIGURE 3. PET-SCAN imaging. Hiper signal captation corroborating with MRI lesions and aditional adenopathy better visualized.

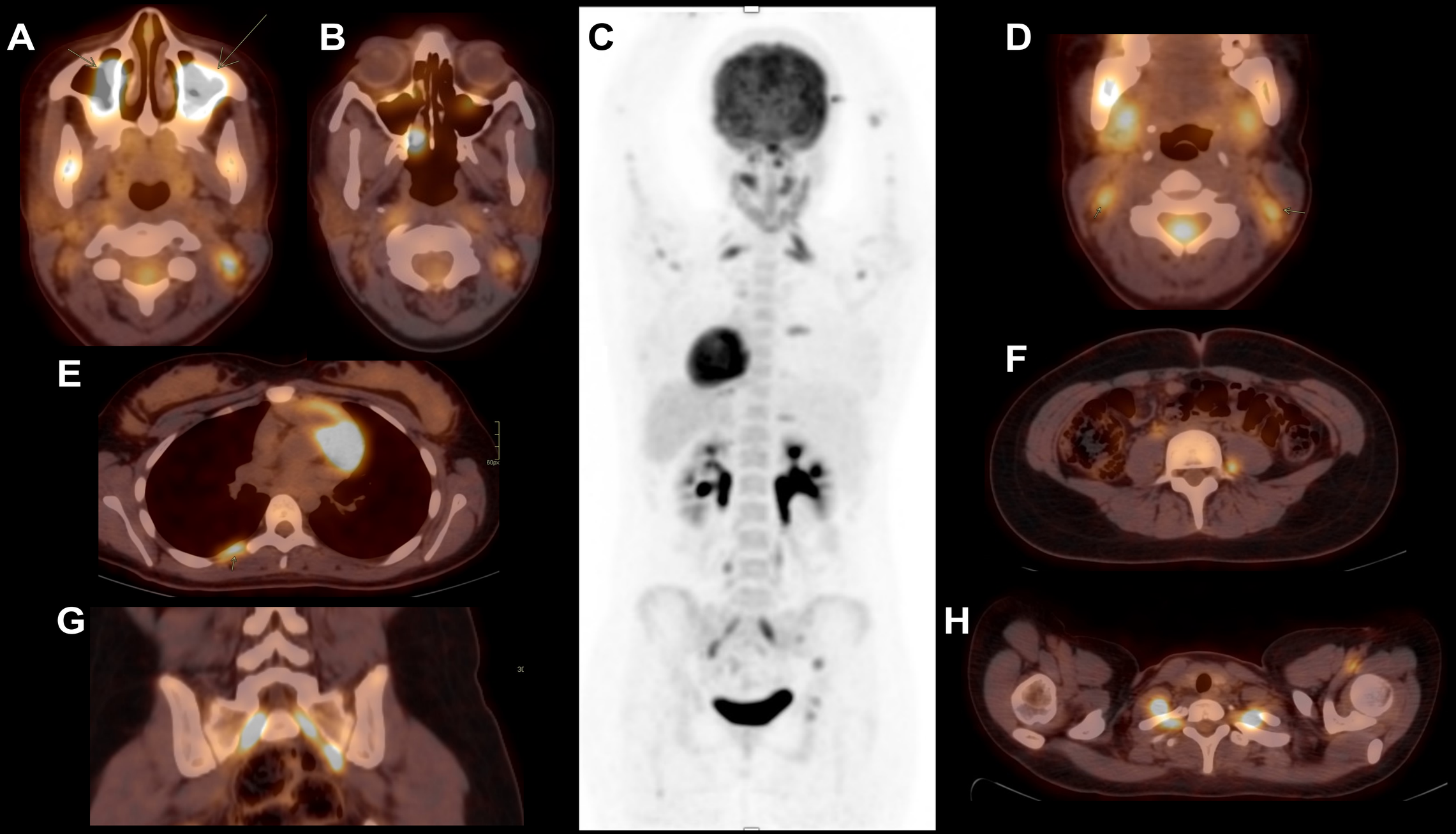
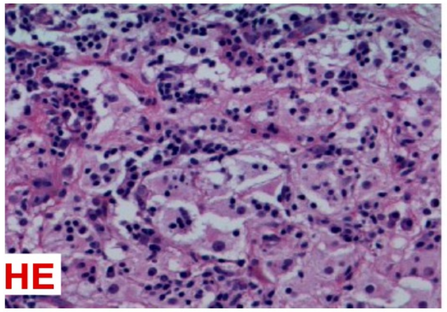
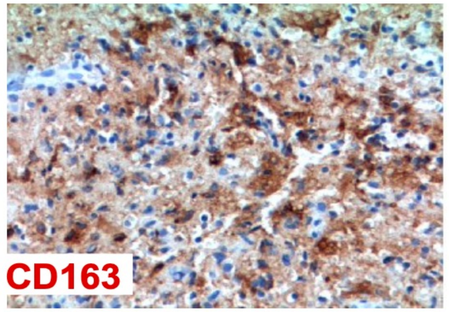
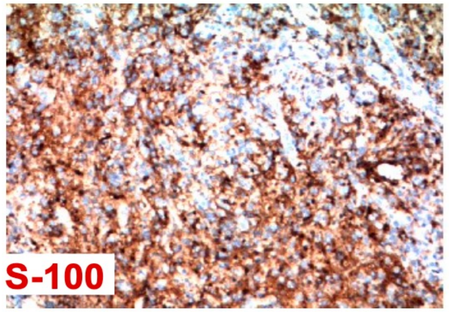


Figure 4: Paranasal sinuses biopsy tissue. Immunohistochemistry (IHC) markers (+CD163/+S-100) and emperipolesis presence on hematoxylin/eosin staining (HE).







Anticorpos	Clone	Resultado	Observação
• CD1a - antígeno de células de Langerhans	O10	Negativo	
• Proteína S-100	Policlonal	Positivo	
• CD207 (Langerina), receptor de lectina tipo C, clone 1206	12D6	Negativo	
• CD163, complexo hemoglobina-haptoglobina	MRQ-26	Positivo	

RESULTS: We describe a rare presentation of RDD as a bilateral choroidal mass in a young patient with vision and hearing loss, in addition to meningeal signs. Clinical symptoms is variable and involve multidisciplinary interpretation. Definitive diagnosis might be made on histology (fig 4). Rosai-Dorfman-Destombes was a challenging diagnostic and should be considered in the differential diagnosis of choroidal metastasis, tuberculosis, lymphoproliferative diseases and other autoimune-inflammatory disorder (e.g. VKH).

DISCUSSION: Sinus histiocytosis with massive adenopathies, also called Rosai-Dorfman-Destombes (RDD) disease, was first described by Paul Destombes in 1965 (1) and Rosai and Dorfman in 1969 (2). RDD is a non-Langerhans cell histiocytosis characterized by accumulation of activated histiocytes, presenting as nodal, or extranodal disease within affected tissues, often bilateral involvement at the same time. RDD occurs in young patients, most characterized by painless cervical lymphadenopathy and fever (3). Extranodal involvement are approximately 43% of cases of RDD, wich most common sites are the nasal cavity and paranasal sinuses, respiratory tract, skin, and central nervous system (4). Orbital cases are almost 10% of RDD, but orbital involvement alone is rare. When the eyes are affected, it usually manifests as an orbital mass, the choroidal mass being very infrequent (5). RDD etiology is still unclear but is also known that it can occur as an isolated disorder or in association with other lymphoproliferative diseases, such as lymphoma.(6) We report a case of atypical presentation of RDD disease with predominance of choroidal and neurological involvement.

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