Erdheim-Chester disease is a non-Langerhans histiocytosis.

Until 2014 at least 550 cases have been reported. According to European Rare Disease Organization and National Organization for Rare Disorders it is a rare disease. The most common symptom is bone pain in the lower extremities and it usually appears between the 5th and 7th decades of life. The diagnostic is based on immunohistochemical results: S100(+/-), CD68(+), and CD1a(-), the latter 2 are mandatory. The best treatment nowadays is alpha-interferon or pegylated alpha-2. The overall survival is 96% at one year and 68% at 5 years. Central nervous system involvement is associated with a worse outcome.

Case series

Valentini et al., report a case of Erdheim-Chester disease and review 60 cases from the literature. These cases are consider to have Erdheim-Chester disease when they have either typical bone radiographs (symmetrical long bones osteosclerosis) and/or histologic criteria disclosing histiocytic infiltration with distinctive immunohistochemical phenotype of the non-Langerhans cell histiocytes with positive staining for CD68 and negative staining for S-100 protein and CD1a. Our patient undergoes chemiotherapy according to the LCH-II stratification and therapy plan (Vinblastine, Etoposide and Prednisone) and thereafter receives Carboplatin and Etoposide, and Somatostatin. She is alive and clinically well 33 months after onset of symptoms and the lesions don't appear to progress at imaging examinations. In conclusion, Erdheim-Chester disease may be confused with Langerhans cell histiocytosis as it sometimes shares the same clinical (exophthalmos, diabetes insipidus) or radiologic (osteolytic lesions) findings. However, the characteristics radiological pattern of Erdheim-Chester disease together the immunohistochemical phenotype of hystiocytic infiltration supports the theory that Erdheim-Chester disease is a unique disease entity distinct [1].

Case reports

An 11-year-old boy, who was diagnosed with LCH 7 years previously, presented with multiple giant intracranial lesions. At the time of his initial diagnosis, only one intracranial lesion was observed, and it began to enlarge. Currently, up to 7 intracranial lesions can be observed in this patient. However, the diagnosis of ECD was not confirmed until this most recent open resection. The BRAF V600E mutation was detected in both LCH and ECD lesions. Dabrafenib therapy exhibited dramatic efficacy in this pediatric patient. This case represents the first successful application of dabrafenib in a pediatric patient with intracranial ECD lesions as well as mixed ECD and LCH. In this article, the authors describe the intricate diagnosis and treatment processes in this patient. Recent studies regarding treatment with BRAF inhibitors for neurological involvement in mixed ECD and LCH are also reviewed [2]

Two cases are presentedwith central nervous system lesions in the absence of lesions in other organs on their onset. Very few cases have been reported with this kind of presentation. We also noted that these patients had recurrences or new lesions at 8 months. A follow-up is proposed with brain MRI and thoraco-abdominal PET every 3-4 months [3]

1)
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2)
Hao X, Feng R, Bi Y, Liu Y, Li C, Lu T, Tian Y. Dramatic efficacy of dabrafenib in Erdheim-Chester disease (ECD): a pediatric patient with multiple large intracranial ECD lesions hidden by refractory Langerhans cell histiocytosis. J Neurosurg Pediatr. 2018 Sep 28:1-6. doi: 10.3171/2018.6.PEDS17728. [Epub ahead of print] PubMed PMID: 30265230.
3)
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