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Hematological aspects of Gaucher's disease in Algeria: About 15 cases

Background: Gaucher's disease (GD) is the most common group of lysosomal storage disorders caused by defective activity of an enzyme β-glucosidase leading to accumulation of glucocerebroside in cells of macrophage lineage. Accumulation of glucosylceramide in tissues leads to multisystem organ involvement. Hematological manifestations of Gaucher's disease represent the most common mode of disease revelation. A diagnostic delay of several years is often a source of serious complications, especially hematological and bone involvement. The hematologist plays an important role in the diagnosis of GD through the reading of the medullogram, which can sometimes be difficult (pseudo-gaucher).

Objective: The main objective of this study is to report the hematological aspects of Gaucher's disease: Frequency, phenotypes, poor outcomes factors and evolution.

Patients & Methods: Our study is a retrospective descriptive study in patients with type 1 GD confirmed by a decreased activity of β-glucosidase and the presence of the coding gene mutation, recruited at the internal medicine department from January 2001 to January 2018.

Results: We collected 15 cases of Gaucher's disease, six men and nine women with a mean age of 41.5 years. Nearly 50% of patients had a diagnosis delay and were diagnosed after the age of 30. The clinical profile was variable nevertheless 86% (13 cases) of patients had cytopenia with 100% thrombocytopenia, 50% anemia and 46% leucopenia. Seventy percent of patients had hepatosplenomegaly of which 23% (three cases) had splenectomy in childhood. The bone involvement was present in all our patients distributed as follows: bone pain (50%), bone infarction (21.42%), Erlenmeyer-like bone deformities (14.28%), vertebral compression (14.28%), osteoporosis (57.14%), osteopenia (21.42%) and osteonecrosis (28.57%) involving hips, knees and shoulders. Only one of our patients had a prosthetic replacement (total hip replacement). Splenectomized patients (three cases) had severe complications including bone (osteonecrosis of the femoral heads) and pulmonary arterial hypertension which was the cause of the death of a patient during pregnancy. Also, the diagnostic and therapeutic delay led to severe complications such as severe cytopenia (two cases) with (thrombocytopenia at 20000), deep anemia (HB: 7g/dl), leucopenia (1600 mm) without infectious manifestations and osteonecrosis of femoral heads (three cases). Ferritinemia and chitotriosidase were done in 50% of patients and were frankly high and correlated with the severity of the disease. Eighty-six percent (86%) of patients treated with enzyme replacement treatment (Imiglucerase) have good outcomes after one year with platelet normalization in 60% of cases, normalization of hemoglobin and leucocytes in all patients except two patients with massive splenomegaly. Normalization of bone mineral density was noted in all osteoporotic patients with associated bisphosphonate therapy, only 30% of patients-maintained splenomegaly.

Conclusion: Cytopenia are common and severe in GD patients with bone complications and massive splenomegaly. Splenectomy is a factor of poor prognosis and it is associated with bone complications and pulmonary arterial hypertension. The clinical phenotype of patients remains variable, possibly linked to a particular genotype. Enzyme replacement therapy has significantly improved the quality of life of patients, cytopenia and bone manifestations.

Biography

Berrah Abdelkrim is Professor of Internal Medicine at Algiers Medical School and Head of Internal Medicine Department at University Hospital Bab El Oued. His internship and residency in Internal Medicine were performed at the University Hospital Mustapha Pacha Algiers where he spent several years as Associate Professor. His interests include autoimmune diseases, vascular disease, non-communicable diseases, hypertension and diabetes. He is representative of Algeria in the Maghreb Academy of Sciences, past President of the Algerian Society of Hypertension, past Chairman of the Pensions Commission of Department of Veterans Affairs, Member of the French Society of Internal Medicine and Foreign Correspondent of "La Revue Française de Médecine Interne". He has authored or co-authored over 300 publications. He serves as Principal Investigator or Investigator in many studies (PRAMIAL, ICLPS, LIRA Ramadan, Xalia LEA, Vision). He is member of an advisory board or a scientific board, speaker and also consultant near several laboratories of the pharmaceutical industry.

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