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Gaucher disease: An orphan disease with significant osseous manifestations

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Gaucher disease the most common of the lysosomal storage diseases and is classified in orphan diseases, that comprise rare disorders with prevalence of 1:50000 or lower in the general population. Gaucher disease results from mutations leading to impaired enzymatic activity of a lysosomal hydrolase called β-glucocerebrosidase and thus to the accumulation of glucocerebrosides in the lysosomes of the macrophages. This has as a result cytopenias due to hypersplenism and infiltration of bone marrow by Gaucher cells. Disease severity varies greatly from the invariably mortal infantile type 2 and the completely asymptomatic type 1. Clinical manifestations include splenomegaly, hepatomegaly and growth retardation. There are three types of Gaucher disease, type 1, 2 and 3. Type 1 accounts for the 95% of cases in patients of Caucasian origin. Also, the activated macrophages excrete cytokines that affect the bones. Osteopenia, osteoporosis, painful bone crises, pathologic fractures, osteonecrosis may occur. In general, skeletal involvement is considered a sign of grave prognosis since it can lead to serious complications with elevated morbidity and mortality. There is significant consideration that disease clinical phenotype should be considered as a continuum and not as discrete clinical subtypes. Early diagnosis of the disease is crucial since most patients have significant splanchnic involvement at the time of diagnosis in types 1 and 3. The major diagnostic criterion is reduced enzymatic activity of β-glucocerebrosidase. Chitotriosidase levels and Chemokine CC (CCL18/PARC) are also measured. Therapy consists of β-glucocerebrosidase substitution and substrate reduction therapy.

Biography

Efstathios Konstantinou Koutsostathis has completed his PhD in Medicine from the National and Kapodostrian University of Athens. He has completed his education in Internal Medicine at Attikon University Hospital. He is an internist, consultant at Kerameikos Health Center. He is also a Post Graduate Student at the Medical School of Athens in the field of Metabolic Bone diseases and in Public Health at the Natonal School of Public Health. He has published papers in medical journals.

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