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Gaucher disease - current challenges in diagnosis and management

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Gaucher disease (GD) is an autosomal recessive disease resulting from mutations in the glucocerebrosidase gene (GBA1) causing decreased activity of the lysosomal enzyme beta-glucocerebrosidase, and therefore leading to accumulation of substrate. It is one of the two most common lysosomal storage disorders, with an overall prevalence of about 1 in 50,000, but with a particular high prevalence among Ashkenazi Jews (~1:800). More than 860 different mutations have been identified in the GBA1 gene, explaining in part the great phenotypic heterogeneity of GD, that goes beyond the 3 clinical forms, which are based on either the absence (type 1; GD1) or presence (types 2 and 3) of neurological signs. Since treatment is only available for type 1 but not to any of the neuronopathic forms, keeping this traditional classification, with ramifications for genetic counselling, is recommended. GD is important for hematologists because the presenting symptoms are typically hematological (splenomegaly, anaemia, thrombocytopenia), because hyperferritinaemia and gammopathies are common, because the diagnosis is frequently made by finding Gaucher cells in bone-marrow aspiration (albeit not the best diagnostic test), because of the need for hematological consultations before surgery and delivery (with platelet dysfunction adding to the risk of bleeding), and finally because of the increased incidence of haematological malignancies (particularly multiple myeloma) and ITP. Recent ex-vivo gene therapy trials involve autologous HSCT, further engaging haematologists with GD. The availability of several therapeutic options, three or more intravenous enzymes (similar but not biosimilars) and 2 different oral formulations for substrate inhibition, may be a mixed blessing if the less suitable drug is prescribed. Also important are the indications for initiation of treatment, as not all patients require specific therapy. My personal approach to the management of patients with GD will be delineated based on 3 decades of experience in the world largest clinic for GD.

Biography

Ari Zimran was the Founder and the Director of the Gaucher Unit at Shaare Zedek Medical Center in Jerusalem, Israel from 1990 to 2018, and is currently a senior physician at the Unit. This is the world's largest referral centre for Gaucher disease (GD), where more than 850 patients have been followed, and about 350 patients are treated with enzyme replacement therapy. Professor Zimran has published more than 320 professional papers and reviews and has edited three books; he has been a leader in clinical trials for new treatments for GD, including Cerezyme™, Zavesca™, VPRIV™, Elelyso™ and Cerdelga™. Professor Zimran was the first to report the possible association between GD and Parkinson disease (PD), and his current research activities include prodromal features of PD (with the ultimate goal of prevention) among GBA carriers, gene therapy and oral delivery of recombinant human proteins.

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