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Gaucher Kayıt Sistemi hastalığın klinik spektrumunun ve spesifik tedavilerinin etkilerinin kapsamlı bir şekilde değerlendirilmesine olanak sağlamaktadır.

Charrow J et al. The Gaucher registry: demographics and disease characteristics of 1698 patients with Gaucher disease. Arch Int Med 2000:160:2835-43.

Background: The Gaucher Registry, the largest database of patients with Gaucher disease (GD) worldwide, was initiated to better delineate the progressive nature of the disorder and determine optimal therapy. This report describes the demographic and clinical characteristics of 1698 patients with GD before they received enzyme replacement therapy.

Methods: Physicians worldwide who treat patients with GD were invited to submit prospective and retrospective data for an ongoing registry, using standardized data collection forms, for central processing and review.

Results: Most patients were from the United States (49%) and Israel (17%), but patients are from 38 countries. Most (94%) had type 1 GD, fewer than 1% had type 2, and 5% had type 3. Mutant allele frequency data, available for 45% of patients, showed the most common alleles to be N370S (53%), L444P (18%), 84GG (7%), and IVS2+1 (2%). Twenty-five percent of L444P homozygotes (13 of 52 patients) had type 1 GD phenotype. Mean age at diagnosis in patients with the N370S/N370S genotype was 27.7 years (SD, 19.7 years); in L444P/L444P patients, 23 years (SD, 3.2 years). Histories of bone pain and radiological bone disease were reported by 63% and 94% of patients, respectively; both were more likely in asplenic patients than in patients with spleens. Mean spleen and liver volumes were 19.8 and 2.0 multiples of normal, respectively. Anemia and thrombocytopenia were present in 64% and 56%, respectively. Thrombocytopenia was present in 13% of asplenic patients.

Conclusions: The Gaucher Registry permits a comprehensive understanding of the clinical spectrum of GD because of the uniquely large sample size. The Registry will be useful in evaluating the effects of specific therapies in GD and the possible influences of environment, ethnicity, and genotype on the natural history of the disorder.

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MAT-TR-2501519