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Gaucher ve ciddi konjenital nötropenisi olan hastada ERT, klinik seyrin çok ağır olmasını engellemiştir.

Kose MD et al. Coexistence of Gaucher Disease and severe congenital neutropenia. Blood Cells Mol Dis 2019;76:1-6.

Gaucher Disease (GD) is the most common lysosomal storage disorder has traditionally been classified into three clinical phenotypes. Type 3 GD is characterized by neurological involvement but neurological symptoms generally appear later in life than in type 2 disease. Neutropenia is much rarer than other hematological manifestations in GD and has not been scrutinized adequately. Severe congenital neutropenia (SCN) is a rare disease entity which is characterized by a paucity of peripherally circulating neutrophils with arrest of neutrophil maturation at the promyelocyte stage and consequent increased susceptibility to severe and recurrent infections. We report a patient who presented in the first year of life with visceral involvement and severe neutropenia in whom the propositus had a unique coexistence of Gaucher Disease and severe congenital neutropenia associated with a mutation in HAX1. In contrast to his expired siblings he had experienced no severe infections. These clinical observations suggest that enzyme replacement therapy may display a modulating factor with respect to the clinical course of SCN.

Synopsis: Our patient is the only report of the combination of Gaucher Disease and Kostmann Syndrome in the literature. The clinical course of our patient is not severe when comparing with exitus siblings and other Kostmann Syndrome patients. But when considering the patient's only clinical difference is ERT, this case is very important to emphasise the role of enzyme replacement therapy in bone marrow.

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