Cerezyme® 30 YIL

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ERT kesildikten sonra hastalık durumunda dramatik regresyon ortaya çıkmaktadır.

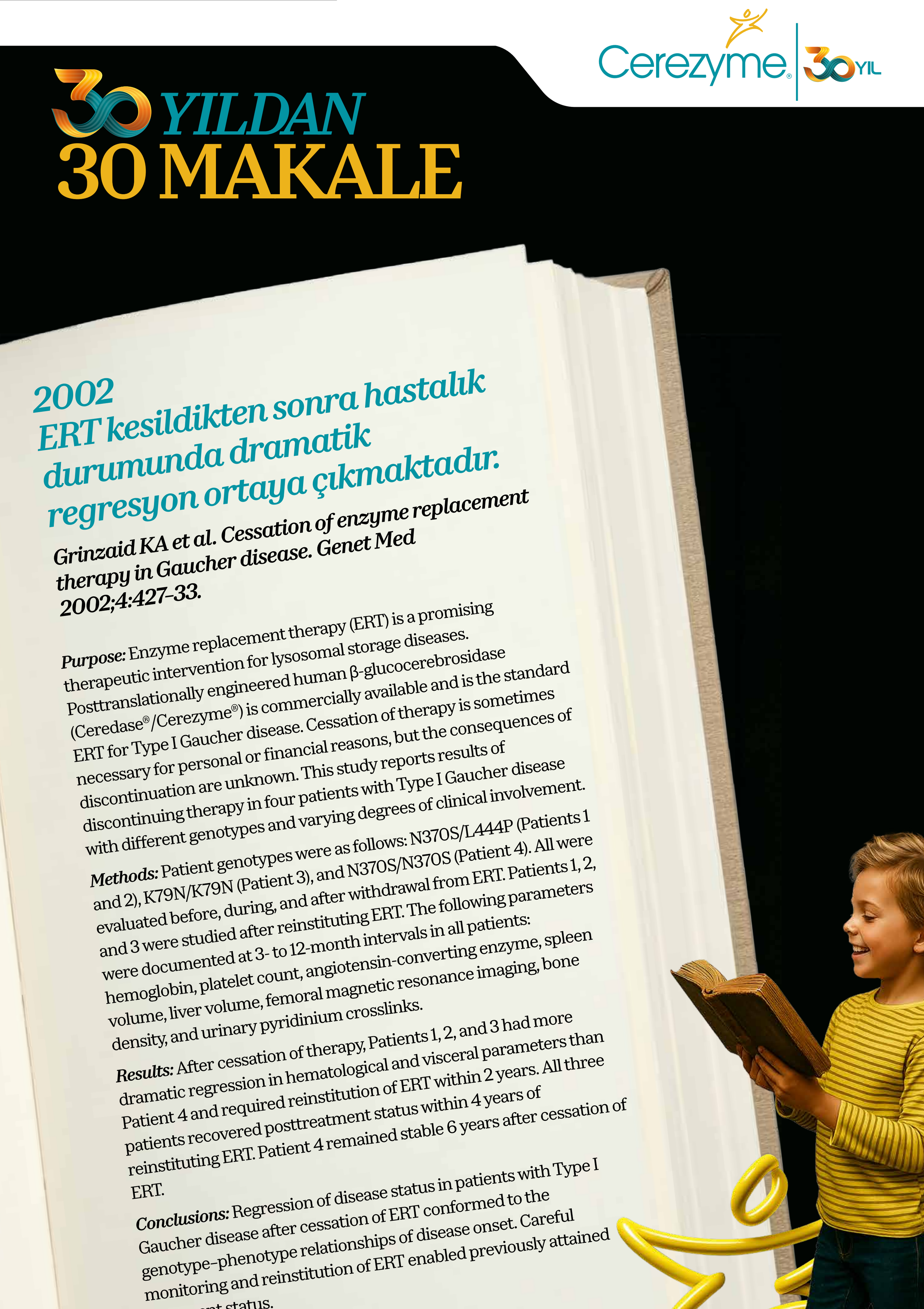
Grinzaid KA et al. Cessation of enzyme replacement therapy in Gaucher disease. Genet Med 2002;4:427–33.

Purpose: Enzyme replacement therapy (ERT) is a promising therapeutic intervention for lysosomal storage diseases. Posttranslationally engineered human β-glucocerebrosidase (Ceredase®/Cerezyme®) is commercially available and is the standard ERT for Type I Gaucher disease. Cessation of therapy is sometimes necessary for personal or financial reasons, but the consequences of discontinuation are unknown. This study reports results of discontinuing therapy in four patients with Type I Gaucher disease with different genotypes and varying degrees of clinical involvement.

Methods: Patient genotypes were as follows: N370S/L444P (Patients 1 and 2), K79N/K79N (Patient 3), and N370S/N370S (Patient 4). All were evaluated before, during, and after withdrawal from ERT. Patients 1, 2, and 3 were studied after reinstituting ERT. The following parameters were documented at 3- to 12-month intervals in all patients: hemoglobin, platelet count, angiotensin-converting enzyme, spleen volume, liver volume, femoral magnetic resonance imaging, bone density, and urinary pyridinium crosslinks.

Results: After cessation of therapy, Patients 1, 2, and 3 had more dramatic regression in hematological and visceral parameters than Patient 4 and required reinstitution of ERT within 2 years. All three patients recovered posttreatment status within 4 years of reinstituting ERT. Patient 4 remained stable 6 years after cessation of ERT.

Conclusions: Regression of disease status in patients with Type I Gaucher disease after cessation of ERT conformed to the genotype–phenotype relationships of disease onset. Careful monitoring and reinstitution of ERT enabled previously attained treatment status.



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