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Gaucher's Disease and Hurler's Syndrome in Two First Cousins

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ABSTRACT Lysosomal storage diseases (LSD) are a group of approximately 50 autosomal recessive inborn errors of metabolism resulting from defects in lysosomal function. These diseases are progressive and multisystemic. The researchers' present two patients from Oaxaca, Mexico, and describe their findings from clinical evaluations, blood biometry, urinalysis, bone-marrow smears, X-rays, enzymatic measurements, and molecular studies. Patient 1, a 10- year-old school-age female with Gaucher's disease (sphingolipidosis) shows two previously unreported mutations in the glucosylceramide beta (GBA) gene. A homozygous mutation in exon 5 (c.463T>C; p.Tyr155His) and a heterozygous variant in exon 10 (c.1459C>A; p.Ala487Thr) were identified. Patient 2, a 3-year-old female suffering from Hurler's syndrome mucopolysaccharidosis type I due to a homozygous 12bp deletion from nucleotides 46 to 57 in the alpha-L-iduronidase (IDUA) gene. In summary, two first-degree Mexican patients with different forms of LSDs, and two previously unreported mutations in the GBA gene are described in this study.