Imiglucerase-Treated Italian Patients with Gaucher Disease Type 1 or Type 3 had the following Long-term Bone Outcomes: A Sub-study from the Global Cooperative Gaucher Gathering (ICGG) Gaucher Vault

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Abstract

A rare condition known to cause skeletal symptoms is Gaucher disease. It can progress to aseptic bone necrosis and pathological fractures in the advanced stage. Although enzymatic replacement therapy (ERT) has significantly improved a patient's quality of life, it has not prevented complications related to the bone. There are very few publications in the literature that have discussed the surgical management of this disorder. The way these patients are handled in orthopedic surgery is very specific.

Most patients with Gaucher illness have moderate and frequently debilitating skeletal indications. The phenotypic diversity of Gaucher disease is well-known, and there are no consistent genotype-phenotype correlations. In Argentina, a public cooperative gathering, Grupo Argentino de Diagnóstico tratamiento de la enfermedad de Gaucher, GADTEG portrayed consistently extreme sort Gaucher illness signs giving in youth a huge weight of irreversible skeletal sickness. Here utilizing Long-Read Single Atom Ongoing (SMRT) Sequencing of the GBA locus, we show that the RecNciIallele is profoundly common and is related to serious skeletal appearances with beginning in adolescence or in youthful grown-ups. In addition, we described a novel, previously unknown GBA variants.

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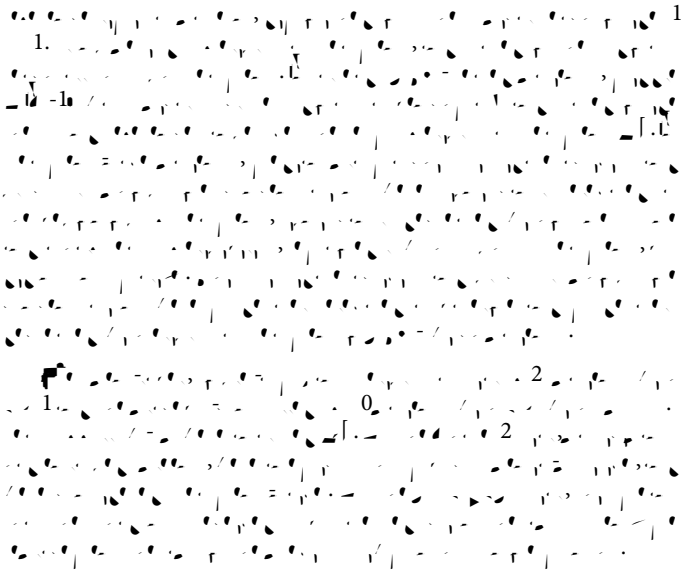

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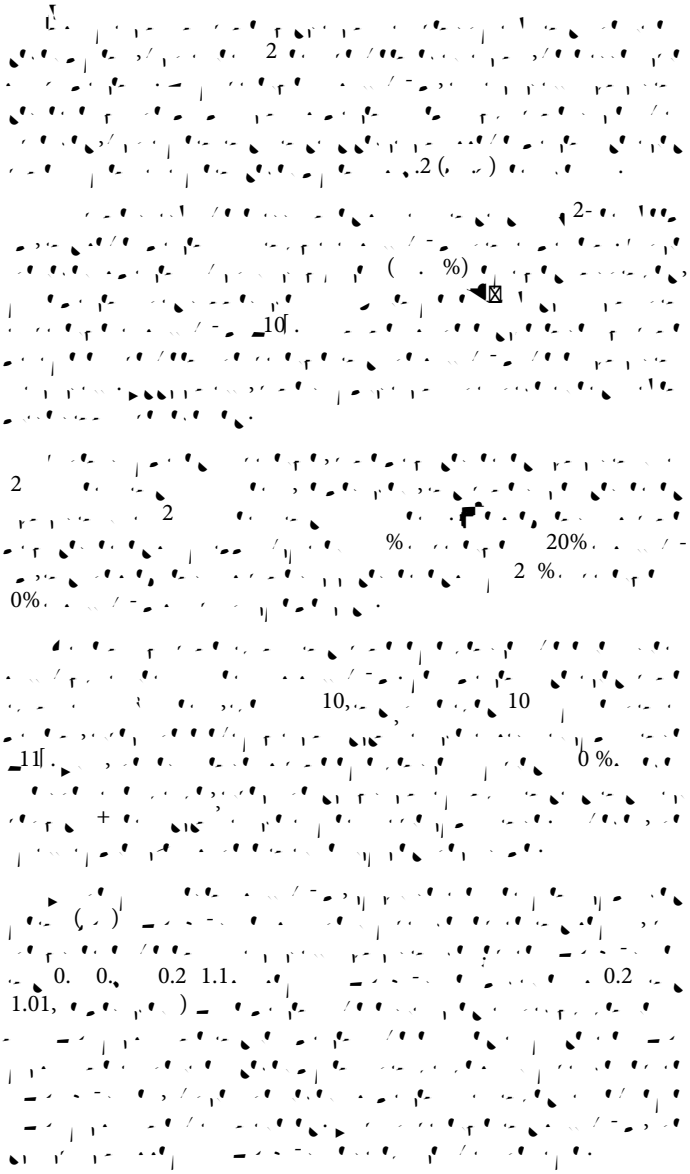

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