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Mucolipidosis II/III in Korea

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Mucolipidosis (ML) II alpha/beta and III alpha/beta are autosomal recessive inherited diseases caused by a dysfunction of the lysosomal enzyme N-acetylglucosamine-1-phosphotransferase (GlcNac-1-phophotransferase). GlcNAc-1-phosphotransferase is involved in the process of attaching a molecule called mannose-6-phosphate (M6P) to specific digestive enzymes. With dysfunction of this enzyme, digestive enzymes cannot be tagged with M6P and transported to lysosomes. The lack of digestive enzymes within lysosomes causes large molecules to accumulate there. We reviewed the medical charts of 12 Korean patients who were diagnosed with ML II/III at Samsung Medical Center. The diagnosis of ML was based on clinical manifestations and lysosomal enzyme activities in serum, lymphocyte and skin fibroblasts. Most patients showed coarse face, developmental delay, multiple joint contracture and skeletal dysplasia with cardiac involvement at the time of diagnosis. There was no significant difference of organomegaly or cardiac involvement between ML II and III patient. However, the patients with ML II showed severe respiratory that need respiratory support and cardiac problems such as hypertrophic cardiomyopathy. We sequenced the GNPTAB gene, which encoded alpha and/or beta subunits of GlcNac-1-phophotransferase, in 4 patients with ML II and 5 patients w with ML III patients. We detected 14 mutations in the GNPTAB gene of the 16 alleles, and most of them were nonsense or frameshift mutation, and only a missense mutation was found. All patients with ML II had compound heterozygotes of nonsense or frameshift mutations. One heterozygous mutation, c.3565C>T (p.R1189X), was found in one patient with ML III and two patients with ML II. This study demonstrates clinical features and molecular analysis in Korean patients with ML II and III.

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