

Mucopolidosis I/II/III in Taiwan

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The mucopolidoses (ML) are a group of rare autosomal recessive lysosomal storage diseases with genetic defects in which their bodies either do not produce enough enzymes or, in some instances, produce ineffective forms of enzymes. In ML, abnormal amounts of carbohydrates and lipids accumulate in cells of various tissues in the body, leading to symptoms that range from mild learning disabilities to severe intellectual impairment and skeletal deformities. ML includes four diseases as follows: ML-I (sialidosis), ML-II (inclusion-cell, or I-cell, disease), ML-III (pseudo-Hurler polydystrophy), and ML-IV. Lai et al.¹⁾ performed a serial analysis of 17 Taiwanese patients with ML-I focusing on evolution of clinical features, electrophysiological findings, genetic studies, and neuroimage examinations. They reported that all patients had a mutation at c.554A→G in exon 3 of the *NEU1* gene causing Ser182Gly substitution. Fifteen patients were homozygous. Two patients were heterozygous with novel mutations, c.956C→T causing Ala319Val in one and c.163C→T causing Gln55stop codon in the other. The neuraminidase activity was markedly decreased in all 11 available patients. Only three patients (17.6%) manifested the macular cherry-red spot. The majority of patients (82.3%) developed full-blown manifestation of myoclonus, ataxia, and seizures within 5 years. Abnormal somatosensory evoked potentials with giant cortical waves were found in all patients. Prolonged P100 peak latency of the visual evoked potentials (VEPs) were found in 16 patients (94.1%) in the early stage even without visual symptoms. ML-I in Taiwanese population illustrates distinct characteristics of phenotype with infrequent cherry-red spot. They suggested screen the *NEU1* mutations in patients presenting action myoclonus with abnor-

mal VEPs, even without macular cherry-red spots. Ma et al.²⁾ reported on the segregation of two *GNPTAB* truncated mutations in a Taiwanese family, causing ML-II phenotype in an infant with an uncommon sign of marked hair color change. Chen et al.³⁾ described the characteristic findings of sonography and magnetic resonance imaging (MRI) of claw hand deformity of a 16-year-old Taiwanese boy with ML-III. The diagnosis of ML-III was confirmed by the presence of elevated activities of β -glucuronidase (2,141.99 nmol/mg protein/hour), arylsulfatase A (1,237.7 nmol/mg protein/hour) and α -fucosidase (52.95 nmol/mg protein/hour) in his plasma and decreased activity of these lysosomal enzymes in cultured skin fibroblasts. They found that sonography and MRI screening for claw hand deformity may offer important clues enabling early diagnosis of ML-III.

References

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