



## Clinical Practice and Therapeutics

## Biopsychosocial issues of a patient with mucopolidosis type II

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Mucopolidosis type II (ML II; MIM: 252500) is an extremely rare lysosomal storage disease caused by homozygous or compound heterozygous mutation in the *GNPTAB* gene over chromosome 12q23.2. Only eight patients have been diagnosed with this disease in Taiwan. It is also called I-cell disease because inclusion bodies can be found in cultures of fibroblasts from these patients. A defect in the enzyme N-acetylglucosaminyl-1-phosphotransferase leads to progressive generalized accumulation of mucopolysaccharides and lipids. In addition to physical disability, symptom-related psychosocial problems at different developmental milestones should be addressed.

The only case found in eastern Taiwan was in a 7-year, 11-month-old boy, who presented with progressive limitation of movement in the small joints since the age of 3 years. The boy had a short stature [body height: 107.7 cm (−3.7 SD), body weight: 19.5 kg (10–25%)], coarse facial features, puffy eyelids, a flat nasal bridge, gingival hypertrophy, malocclusion, a protruding abdomen, claw hands, and limited range of motion of multiple joints, but no umbilical hernia; his liver and spleen could not be palpated (Fig. 1). A panel of enzyme assays revealed marked increases in most lysosomal enzymes such as beta-glucuronidase of 1333.33 nmol substrate/h/mL (normal range: 5.6–101.5 nmol substrate/h/mL)

and beta-hexosaminidase of 9600 nmol substrate/h/mL (normal range: 228–1123 nmol substrate/h/mL). Gene testing revealed compound heterozygous mutations of 1090 C→T (R364X) and 1209 T→C (I403I) over the *GNPTAB* gene.

The patient's hearing was normal. Eye examination revealed mild bilateral corneal clouding and mild myopia (visual acuity: 0.6). Very mild mitral valve regurgitation and mild scoliosis, and grade 1 anterior spondylolisthesis of the lumbar spine were noted on cardiac sonography and radiography, respectively.

After entering elementary school, the patient presented multiple somatic complaints including chest pain, difficulty breathing, and bad dreams. He also had many psychosocial difficulties such as poor frustration tolerance, a short attention span, quick loss of temper, and difficulty with impulse control; was easily distracted during learning; and presented with compensatory behaviors when he perceived himself to be inferior to his peers. The school nurse and teachers described the patient as “a small man with high ambition” characterized by much activity, showing off, dangerous behaviors, and physical accidents in school. Along with self-blame, the parents were overindulging, and did not seem to have a plan to cope with these difficulties.

To deal with these problems, the patient had regular visits to a child psychologist to build self-esteem, improve his impulsive behavior and negative mood, and intervention with social adaptation skill training. The patient had restricted fine motor skills and a low average range of intelligence on the Wechsler Preschool and Primary Scale of Intelligence-Revised (full IQ 82, performance IQ 85,

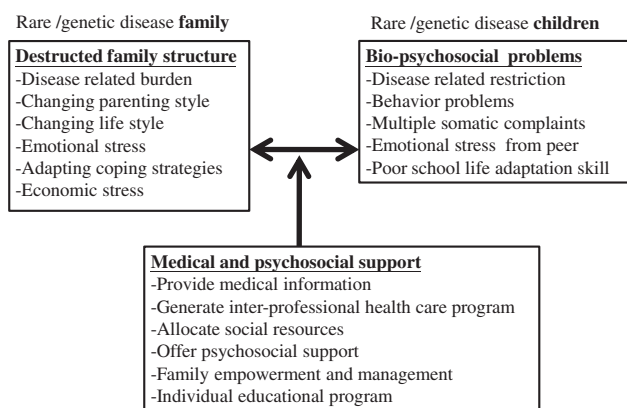
Conflicts of interest: none.

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**Fig. 1.** Patient with mucopolidosis type II. (A) Coarse facial features, depressed nasal bridge, puffy eyelids, widely spaced teeth, thick skin, and claw hands. (B) Multiple subcutaneous nodules around the right ankle joint. (C) Limited range of shoulder motion. *Note.* Published with permission from the patient and his parents.



**Fig. 2.** Supporting system for families and children with genetic/rare diseases.

and verbal IQ 82). His relative strengths (vocabulary, comprehension, object assembly, and geometric design) and weaknesses (arithmetic and similarities) needed to be addressed. A partial individual educational program was arranged.

Empowerment for school teachers and nurses to enhance their knowledge and caring techniques for students with ML II was important. Psychosocial support for the parents and remodeling of parenting were also suggested (Fig. 2). Symptom-related psychiatric

disorders and psychosocial adjustment problems are important issues for family and children affected by rare genetic diseases. A therapeutic daily-life correlation program should be offered for every individual patient.

#### Further reading

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