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Psychosocial perspectives of a patient with mucopolysaccharidosis type VI

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The mucopolysaccharidoses (MPS) are a group of rare inherited lysosomal storage disorders. One subtype of type VI MPS, also known as Maroteaux–Lamy syndrome, is characterized by systemic clinical manifestations with symptoms related to the skeletal, cardiac, respiratory, ophthalmologic, and central and peripheral nervous systems.

A 21-year-old female college student was diagnosed with MPS type VI at 3 years of age and received weekly naglazyme replacement therapy since she was 12 years old (Fig. 1). Thoracolumbar spine radiography revealed flattening of the vertebral bodies, lumbar flattening of the vertebral bodies, lumbar scoliosis with bilateral dysplastic hips, and deformity of the femoral heads (Fig. 2). Spinal stenosis of the second and third cervical vertebrae with cervical cord compression and myelomalacia were noted on head and cervical magnetic resonance imaging and at recent clinical follow-up. Integrated care has been initiated but the main focus was on the diverse spectrum of disease manifestations.

In order to offer more holistic clinical care, the patient's psychosocial status was assessed by a set of semistructured questions, mixed with open-ended and closed questions focused on self-identity, coping strategies for school life and the disease effects, the patient's vision of the meaning of life, and the expected roles of friends and medical personnel. A 1-hour interview was scheduled

Fig. 1. Photograph of the patient. Note the coarse facial features, large head, frontal bossing, thick eyebrows, depressed nasal bridge, full lips, short stature, and claw hands. Reproduced with permission from the patient.

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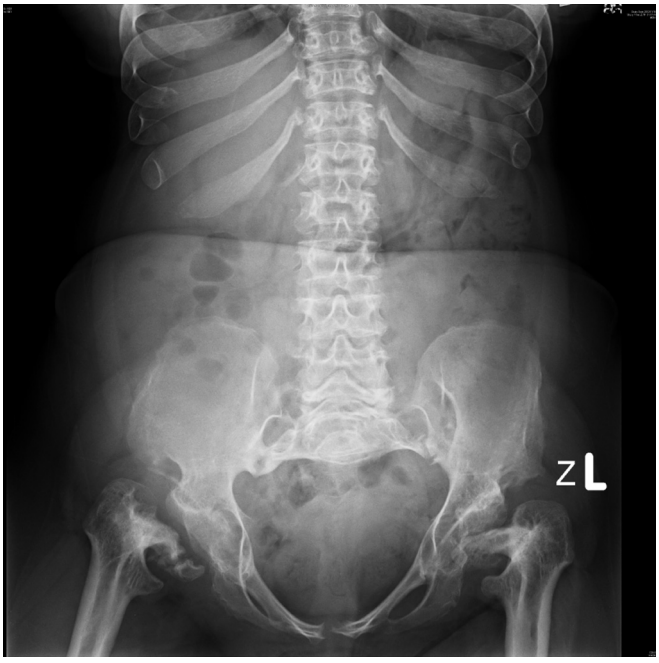

Fig. 2. Thoracolumbar spinal radiograph. Note the oar-shaped ribs (ribs take off from the vertebral bodies and broaden distally) and dysplastic vertebral bodies. Bilateral dislocated hips and deformity of the femoral heads are also observed.

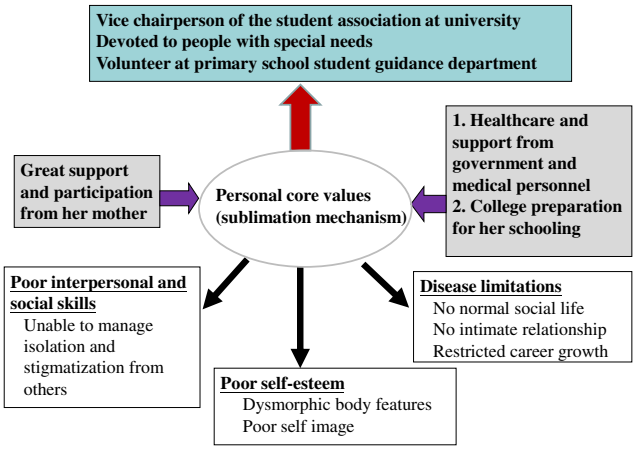

Fig. 3. Results of interview. Black arrows indicate restricted and negative forces noted from patient's life experience. Purple arrows indicate supporting systems identified. Red arrow: sublimation mechanism driving patient holding a positive personal core value for altruistic behavior.

with the patient's permission for recording and publication. Content analysis was done by three researchers and interpretation was by consensus.

Self-awareness of the disease began at the age of 7 years because of her short stature compared with that of her school-aged peers. Most perceptions from schoolmates were negative. She felt intentionally isolated from her peers and most of her teachers at school prior to entering college. She accepted the unfriendly environment silently. Her experience with healthcare professionals was satisfactory. The physicians and medical personnel she encountered were very friendly and supportive. She appreciated participating in a Phase III clinical trial in England and received enzyme replacement therapy free of charge in Taiwan. The patient seemed more emotionally positive with other MPS patients and was an active listener. Her mother was the most important support person in her life.

She is clear about her intentions and understands her situation. She cares about children, and studied subjects dealing with children and the family in college. At school she strives to do her best and tries to play an optimal role in class. She wants to be remembered by society in some special way. She also wants an intimate relationship. Searching for an identity from others is still a lifelong issue for her, and she is afraid of death and refuses to face it (Fig. 3).

With the support of a sublimation mechanism, she holds a positive personal core value for altruistic behavior despite three negative forces from her life experience.

Generation of a friendly growth environment through public education, continued psychological support, and generation of adaptive life skills from early childhood are mandatory for people with special needs.

Further reading

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