Survival and diagnostic age of 175 Taiwanese patients with mucopolysaccharidoses (1985–2019)

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**Background:** Mucopolysaccharidoses (MPSs) are a group of inherited metabolic diseases, which are characterized by the accumulation of glycosaminoglycans, and eventually lead to the progressive damage of various tissues and organs.

**Methods:** An epidemiological study of MPS in Taiwan was performed using multiple sources. The survival and diagnostic age for different types of MPS between 1985 and 2019 were evaluated.

**Results:** Between 1985 and 2019, there were 175 patients diagnosed with MPS disorders in the Taiwanese population, with a median diagnostic age of 3.9 years. There were 21 (12%), 78 (45%), 33 (19%), 32 (18%) and 11 (6%) patients diagnosed with MPS I, II, III, IV and VI, respectively, with median diagnostic ages of 1.5, 3.8, 4.7, 4.5 and 3.7 years, respectively. Diagnosis of MPS patients was significantly earlier in recent decades (p < 0.01). Pilot newborn screening programs for MPS I, II, VI, IVA, and IIB were progressively introduced in Taiwan from 2016, and 48% (16/33) of MPS patients diagnosed between 2016 and 2019 were diagnosed by one of these screening programs, with a median diagnostic age at 0.2 years. For patients born between 2016 and 2019, up to 94% (16/17) were diagnosed with MPS via the newborn screening programs. At the time of this study, 81 patients had passed away with a median age at death of 15.6 years. Age at diagnosis was positively correlated with life expectancy (p < 0.01). Life expectancy also significantly increased between 1985 and 2019, however this increase was gradual (p < 0.01).

**Conclusions:** The life expectancy of Taiwanese patients with MPS has improved in recent decades and patients are being diagnosed earlier. Because of the progressive nature of the disease, early diagnosis by newborn screening programs and timely implementation of early therapeutic interventions may lead to better clinical outcomes.