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Unmet needs of adults living with mucopolysaccharidosis II: data from the Hunter Outcome Survey

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Abstract

Background Mucopolysaccharidosis II (MPS II) is a rare, life-limiting lysosomal storage disease caused by deficient iduronate-2-sulfatase activity. The current standard of care for MPS II is intravenous enzyme replacement therapy (ERT), which has been shown to improve somatic signs and symptoms and to increase life expectancy by approximately 12 years. This study reported on the somatic disease burden and clinical requirements of adult male patients in the Hunter Outcome Survey (ClinicalTrials.gov Identifier: NCT03292887).

Results Of the 373 patients in the analysis, 88 (23.6%) had cognitive impairment and 332 (89.0%) had received ERT. Almost half of all ERT-treated patients (47.0%) had undergone surgery in adulthood; the most common surgery was hernia repair (17.8% of patients). Over one-third (38.6%) reported hearing aid use. The median 6-min walk test distance for 151 treated patients was 436.0 m at the latest assessment after 18 years of age. Cardiovascular signs and symptoms were present in 71.6% (192/268) of patients and 27.3% (60/220) reported oxygen dependency after 18 years of age. Approximately half (50.9%) of ERT-treated patients experienced at least one serious adverse event in adulthood, with the most common being respiratory disorders. Intravenous ERT was well tolerated, with a rate of serious infusion-related reactions in adulthood of 0.03 per 10 patient-years.

Conclusions Overall, adult patients with neuronopathic and non-neuronopathic MPS II had a high disease burden and requirement for surgeries, emphasizing the need to continue multidisciplinary management and regular assessments in adulthood. Further research into the differences in care needs of adult patients with MPS II is warranted.

Trial registration NCT03292887.

Keywords Adults, Cognitive Impairment, Hunter Syndrome, Mucopolysaccharidosis type II, Neuronopathic, Non-neuronopathic

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