J Inherit Metab Dis (2007) 30:821
DOI 10.1007/s10545-007-0551-9

SHORT REPORT

Enzyme replacement therapy in 12 patients with MPS I–H/S with homozygous p.Leu490Pro mutation

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Received: 3 January 2007 / Submitted in revised form: 11 May 2007 / Accepted: 14 May 2007 / Published online: 14 June 2007

Online citation: JIMD Short Report #059 (2007) Online

Summary We describe a cohort of 14 Hurler–Scheie patients homozygous for the p.Leu490Pro missense mutation in the α-L-iduronidase gene. Now based in the UK, they are all of Pakistani/Kashmiri descent; 64% were female; 11/14 (79%) had a sibling or cousin with MPS I and the parents are consanguineous in all cases. The median age at diagnosis was 1.8 years (range from antenatal diagnosis to 16.5 years). Twelve were on ERT with recombinant human α-L-iduronidase (IDUA; Laronidase, Genzyme) for a median duration of 22.5 months (range 2–71 months) and median age at commencement of ERT was 8.6 years (range 0.4–23.1 years). There was clear improvement in the size of liver and spleen as well as reduction in urine glycosaminoglycans (GAGs). The mean (range) urine GAG levels in mg/mmol creatinine were 63.4 (28.9–105.6), 28.3 (10.9–41.4), 22.8 (12.1–43.1), 15.7 (9.2–24.8) and 16.3 (10.1–21.0) at commencement, 3 months post ERT, 6 months post ERT, 12 months post ERT and 24 months post ERT, respectively. Effects on growth were not clear as there does not seem to be an obvious trend of increase or decrease in height after commencement of ERT and this seems to be the case regardless of the age at which ERT was started.

Communicating editor: Irene Marie

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Electronic Supplementary Material
The online version of this article (doi:10.1007/s10545-007-0551-9) contains supplementary material, which is available to authorized users.