IN THIS ISSUE

Pediatric patients tolerate effective enzyme replacement therapy for ASMD

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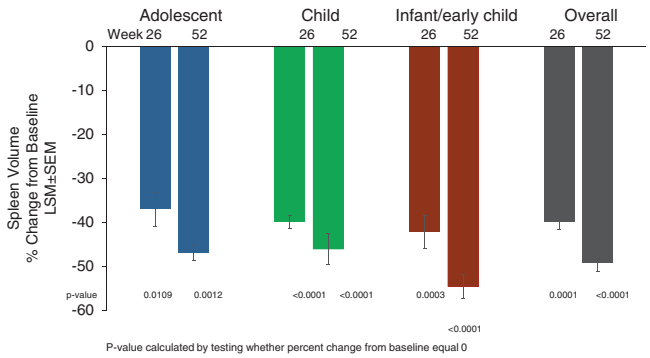

Pediatric patients with chronic acid sphingomyelinase deficiency (ASMD), a condition historically known as Niemann–Pick disease types A/B or B, are at risk for premature death. Sequence variants in *SMPD1*, a gene encoding a lysosomal enzyme, lead to intracellular accumulation of sphingomyelin, an abundant lipid. Sphingomyelin buildup results in hepatosplenomegaly, dyslipidemia, interstitial lung disease, and delayed growth, among other complications. Patient management relies on supportive care. However, in adults, enzyme replacement therapy with olipudase alfa, a recombinant human acid sphingomyelinase that likely does not cross the blood–brain barrier, showed promise for treatment. In this issue, Diaz and colleagues report that olipudase alfa treatment improved disease pathology and was well tolerated in pediatric patients with chronic ASMD. The researchers enrolled 20 patients aged 1.5 to 17.5 years old in the study, all of whom completed the 64-week trial. At baseline, most patients showed severe splenomegaly, and up to a third presented with respiratory disease, excessive bleeding/bruising, and/or thrombocytopenia at disease onset. Patients received olipudase alfa infusions every 2 weeks, starting at a minimal dose and ramping up to the maintenance dosage of 3 mg/kg. Although all patients experienced at least one adverse event, nearly 90% of events were mild. One patient experienced an anaphylactic reaction; however, desensitization enabled the patient to eventually reach the maintenance dose. Treatment reduced patient spleen and liver sizes by more than 40%, improved lung diffusion capacity, and normalized lipid profiles and liver transaminase levels. Most patients also showed marked increases in height. Overall, patients with the most severe disease at the beginning of the study exhibited the greatest improvement. Together, the findings indicate that enzyme replacement therapy with olipudase alfa is a well-tolerated and effective treatment approach for ASMD in children. —*V. L. Dengler, News Editor*

Survey reveals shortage of medical geneticists

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A 2003 survey of the medical genetics workforce revealed a shortage of genetics health-care professionals. More than 15 years later, through the American College of Medical Genetics and Genomics 2019 survey of the medical genetics workforce, Jenkins and colleagues found that the gap between needed genetics services and workforce capacity remains. The researchers analyzed responses to a 32-question, emailed survey from nearly 500 clinical geneticists who are actively seeing patients. The analysis highlighted several trends, including a shortage of medical geneticists, particularly younger and nonwhite professionals. A majority of respondents were female, and nearly 80% were white. A quarter of respondents plan to retire within the next 10 years. The survey also revealed that more than a third of professionals are in just five states, and most are concentrated in urban areas, which limits rural access to services. Although the use of telemedicine doubled—from 16% to 33%—since 2015, the alternative service is insufficient to address the growing needs of patients. New patient visits rose from 6 to 13 per week, and wait times for new emergency and nonemergency patients were often significant. Although the number of clinical genetics residents in residency programs has increased, about a third of approved and funded clinical genetics residency positions remain unfulfilled. Together the survey results highlight the clinical genetics workforce shortage and underscore potential solutions such as increasing workforce recruitment, expanding diversity to better match the US patient population, and enhancing collaborative practice, for which salary augmentation and increased funding support for trainees will be integral, according to the authors. The researchers conclude that successful enhancement of the medical genetics workforce will benefit patients. —*V. L. Dengler, News Editor*

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