

**The long-term outcomes of presymptomatic infants transplanted for Krabbe disease:**

**Report of the workshop held on July 11 and 12, 2008, Holiday Valley, New York**

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**Abstract:** Krabbe disease (globoid cell leukodystrophy) is an autosomal recessive disorder of white matter resulting from deficiency of galactosylceramide beta-galactosidase (GALC) and the consequent accumulation of galactosylceramide and psychosine. Although most patients present within the first 6 months of life, i.e., the early infantile or “classic” phenotype, others present later in life including in adolescence and adulthood. The only available treatment for infants with early infantile Krabbe disease is hematopoietic cell transplantation (HCT), typically using umbilical cord blood. Although transplanted children are far better neurologically than they would have been had they followed the typical fulminant course of early infantile Krabbe disease, anecdotal reports have surfaced suggesting that the majority of presymptomatic children transplanted for Krabbe disease have developed motor and language deterioration. The cause and extent of the deterioration is unknown at this time. With the advent of universal newborn screening for Krabbe disease in New York State and the projected start of screening in Illinois in 2010, understanding the outcome of treatment becomes of paramount importance. Thus, the purpose of this workshop was to bring together child neurologists, geneticists, neurodevelopmental pediatricians, transplanters, neuroradiologists, neurophysiologists, developmental neurobiologists, neuroscientists, and newborn screeners to review the results of the transplantation experience in humans and animals and, if neurologic deterioration was confirmed, develop possible explanations as to causation. This workshop was the first attempt at a multicenter crossdiscipline evaluation of the results of HCT for Krabbe disease. A broad range of individuals participated, including clinicians, academicians, and authorities from the National Institutes of Health, American College of Medical Genetics, and Department of Health and Human Services. *Genet Med* 2009;11(6):000–000.

**Key Words:** *Krabbe disease, globoid cell leukodystrophy, outcomes transplantation*