Recommendation for the Addition of Metachromatic Leukodystrophy (MLD) to the Iowa Newborn Screening Panel

Iowa Health and Human Services Council Ad Hoc Committee for Management of the Newborn Screening Panel

Kimberly Noble Piper

Executive Officer, Center for Congenital and Inherited Disorders

IOWA | Health and Human Services

Metachromatic Leukodystrophy (MLD)

MLD is an inherited condition that causes a defect in a gene that produces an enzyme necessary to reduce the accumulation of toxic sulfides that cause demyelination of the nervous system. Incidence is estimated at 1 in 100,000 births. In Iowa, with a birthrate of ~ 36,700, this would be about one newborn every three years.

Symptoms are progressive and include neuromuscular dysfunction such as weakness, loss of motor skills, and possibly seizures, with progression to vision and hearing loss.

If left undiagnosed and untreated, most newborns with the infantile-onset type of MLD die within 5 years of diagnosis.

Early treatment is imperative before symptoms start. Once neuromuscular dysfunction begins, it cannot be reversed or restored.

MLD Treatments

Treatment for MLD is supportive care for symptoms.

Stem cell transplants to introduce stem cells with functioning genes may improve the disease course in later-onset types of MLD but is less effective for infantile types. Enzyme replacement therapy does not cross the blood/brain barrier, so has no effect on brain or central nervous system function.

Gene therapy is a newer treatment that takes the baby's own stem cells, treats them by introducing copies of a functioning gene, then returns the treated cells back to the baby so the baby's body may start to produce the necessary enzyme.

Ad Hoc Committee Review

The Ad Hoc Committee met in November and December to conduct an evidence-based review of MLD for consideration of adding it to Iowa's newborn screening panel, using an established review framework.

Committee members examined peer-reviewed literature and information from other state newborn screening programs screening for MLD and heard a presentation from an Iowa Board Certified medical geneticist experienced in providing care for children with lysosomal disorders such as MLD. Iowa Newborn Screening Program (INSP) staff spoke to the capacity of the INSP to screen for MLD, and the parents of a child with MLD testified as to their lived experience.

Parallel to this review, Kimberly Noble Piper met with Iowa Medicaid partners to update them on the current newborn screening panel and the potential addition of MLD, including treatment options and the possible cost of treatments.

Ad Hoc Committee Recommendation

After discussion and deliberation, the Ad Hoc Committee reached consensus on a recommendation to add MLD to Iowa's newborn screening panel.

The benefits to newborn screening for MLD were that it allowed early identification of the condition and early treatment before symptoms start, therefore mitigating the impact of the condition to the newborn and family.

If a baby is not screened for MLD, diagnosis of the condition would be based on symptoms that would precipitate a diagnostic workup. By the time symptoms are evident, the condition has already progressed to the point that neuromuscular function has degraded and cannot be restored by treatments.

The Committee puts forth a Level A2 Recommendation:

"Screening for the condition has a high certainty of significant net benefits and screening has high or moderate feasibility. INSP has developmental readiness to screen within 18 months."