Clinic reduces GA1 brain injury risk by 83% with therapies developed over 30 years

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A new study summarizes over 30 years of clinical experience in the treatment and management of glutaric acidemia type 1 (GA1), a rare and potentially devastating metabolic disorder caused by variants in the GCDH gene. The study followed the clinical course of 168 individuals with GA1 who were born between 1973 and 2019 and originated from 26 states and 6 countries. Participants were divided into three cohorts based on timing of diagnosis and method of treatment. The study was a broad collaborative effort led by clinicians and researchers at the Clinic for Special Children (CSC) and will appear in *Molecular Genetics and Metabolism*. It establishes a safe and highly effective standard-of-care for the treatment of GA1, and should serve as a rich and valuable resource for dieticians, physicians, and GA1 families throughout the world for years to come.

Before the CSC's founding in 1989, 90% of infants and young children with GA1 suffered a catastrophic form of acute neurological degeneration. The brain injury of GA1 leaves children mute, wheelchair-dependent, and fully disabled by generalized dystonia, and often results in complications such as scoliosis, hip dislocation, pulmonary aspiration, chronic pain, and untimely death.

Today, with the benefit of early diagnosis, dietary therapy, and an effective hospital protocol, only 7% of children born with GA1 suffer brain injury. Specifically, state newborn screening coupled with strict dietary management reduces the risk of brain injury 14-fold, and uninjured children with GA1 have normal growth, motor development,

and cognitive function. Overall, early diagnosis of GA1 with lysine-free, arginine-enriched metabolic formula and emergency IV infusions during the first two years of life is safe and effective—preventing over 90% of brain injuries. The need for dietary and emergency IV therapies beyond early childhood is uncertain at this time.

More information: Kevin A. Strauss et al, Glutaric acidemia type 1: Treatment and outcome of 168 patients over three decades, *Molecular Genetics and Metabolism* (2020). DOI: 10.1016/j.ymgme.2020.09.007

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