ACD
ASSOCIATION FOR CREATINE DEFICIENCIES

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The X-linked inheritance pattern of Creatine Transporter Deficiency (CTD) creates a challenging situation for estimating the prevalence of this disease. Carrier mothers may unknowingly be mildly impacted by CTD and as a result be less likely to participate in population carrier studies used to estimate prevalence. In addition, unique mutations observed in the CreatineINFO patient registry community (at last review over 80% of mutations were unique) highlights the difficulty of projecting incidence due to frequent de novo mutations that are not captured by carrier studies.

We believe a strong indirect indicator of CTD prevalence is reflected in our global patient community numbers. The CreatineINFO registry has 2.4 times more CTD participants than GAMT participants. The same ratio has been observed in our community support group over the past twelve years. GAMT estimates continue to improve thanks to the adoption of newborn screening and resulting data. In 2022, a scientific evidence review (Kemper, et. al LINK) of the feasibility of GAMT newborn screening calculated a rate of 1:540,276 infants are expected to be born with GAMT. Applying this rate directly to the current world population, we estimate 14,854 GAMT patients worldwide. 2.4 times this projection estimates **35,650 CTD patients** worldwide or a CTD prevalence of **1:225,105**.

We feel this is a conservative estimate. It does not factor in that GAMT may have more clinical awareness and testing than CTD due to newborn screening advocacy efforts and that mildly affected females with CTD are less likely to obtain a diagnosis.

Sincerely,

Heidi Wallis
ACD Executive Director