

address surgical gaps. Barriers included additional stakeholder's perceptions of low user acceptability and appropriateness in some cases and the need for additional study data to inform decision making for practice and policy. **DISCUSSION/SIGNIFICANCE OF IMPACT:** The innovation is efficacious, acceptable, adds to current coping strategies, and fits within existing fistula programs. Stakeholders' pre-implementation perceptions highlight the importance of partnerships and the need for an evidence base related to effectiveness, acceptability, and cost. Challenges to address include access to resources within these contexts (water, soap, and safe space to empty cup) and development of a culturally appropriate counseling message. Future research warranted.

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Developmental Outcomes of Aicardi Goutieres Syndrome

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OBJECTIVES/SPECIFIC AIMS: Metachromatic leukodystrophy (MLD) is a rare, lysosomal storage disorder caused by decreased enzymatic activity of arylsulfatase A. This can be the result of mutations in the ASA gene, or in rare cases PSAP. Historically, MLD has been subdivided into 3 forms based on age of onset: late infantile, juvenile, and adult. These subtypes were defined decades ago, prior to the appreciation of the full clinical spectrum of this lysosomal storage disorder and the advent of genetic testing. As a consequence, these empiric age-based historical definitions do not fully account for the spectrum of disease and are not founded in evidence-based analysis of phenotypic cohorts. Additionally, the antiquated definitions do not fully predict presenting features or disease course, and they fail to stratify outcomes in the few therapies currently available to treat this disease. As novel targeted therapeutics are developed, it is essential to have a clear understanding of the clinical presentation and natural history of MLD. Without properly defined sub-populations, it is difficult to design a therapeutic clinical trial that can demonstrate efficacy in a heterogeneous group. **METHODS/STUDY POPULATION:** In this project, we collected the retrospective natural history of over 50 individuals from around the world. We created an electronic database in REDCap to able to longitudinally collect clinical information. Using this retrospective natural history approach to understanding the disease course of individuals affected by MLD, we were able to characterize age of onset, delay to diagnosis, and common presenting features. **RESULTS/ANTICIPATED RESULTS:** Our results suggest distinct clinical phenotypic subgroups, with distinct presentations. **DISCUSSION/SIGNIFICANCE OF IMPACT:** With a better understanding of the natural history of MLD, we will be able to better counsel families and to design clinical trials with more coherent cohorts and more appropriate clinical endpoints.

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First in Man

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OBJECTIVES/SPECIFIC AIMS: A mimic of congenital infections and a rare genetic cause of interferon overproduction, Aicardi Goutières Syndrome (AGS) results in significant neurologic

disability. AGS is caused by pathogenic changes in the intracellular nucleic acid sensing machinery (TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR1, and IFIH1). All affected individuals exhibit neurologic impairment: from mild spastic paraparesis to severe tetraparesis and global developmental delay. We hypothesize that genotype influences the heterogeneous developmental trajectory found in AGS. **METHODS/STUDY POPULATION:** To characterize this spectrum, age and symptoms at presentation and longitudinal developmental skill acquisition was collected from an international cohort of children (n=88) with genetically confirmed AGS. **RESULTS/ANTICIPATED RESULTS:** We found that individuals present at variable ages, with the largest range in SAMHD1, ADAR, and IFIH1. There are 3 clusters of symptoms at presentation: altered mental status (irritability or lethargy), systemic inflammatory symptoms, and acute neurologic symptoms, with variability across all genotypes. By creating Kaplan-Meier curves for developmental milestones, we were able to create genotype-based developmental trajectories for the children affected by the 5 most common genotypes: TREX1, IFIH1, SAMHD1, ADAR, and RNASEH2B. Individuals with AGS secondary to TREX1 were the most severely affected, significantly less likely to reach milestones compared to the other genotypes, including head control, sitting, and nonspecific mama/dada (p-value <0.005). Individuals affected by SAMHD1, IFIH1, and ADAR collectively attained the most advanced milestones, with 44% of the population achieving a minimum of a single word and 31% able to walk independently. Three retrospective scales were also applied: Gross Motor Function Classification System, Manual Ability Classification Scale, and Communication Function Classification System. Within each genotypic cohort, there was pronounced heterogeneity. **DISCUSSION/SIGNIFICANCE OF IMPACT:** Our results demonstrate the influence of genotype on early development, but also suggest the importance of other unidentified variables. These results underscore the need for deep phenotyping to better characterize subcohorts within the AGS population.

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Healthy eating, physical activity, sleep and cognitive function in elderly population: Data from National Health and Nutrition Examination Survey 2011-2014

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OBJECTIVES/SPECIFIC AIMS: To examine the relationship between healthy eating, physical activity (PA), sleep problem and hours of sleep and cognitive function among elderly population and the racial/ethnic differences in this relation. **METHODS/STUDY POPULATION:** We analyzed data from National Health and Nutrition Examination Survey 2014-2016 for 882 population 60 years and older. Cognitive status was measured by the Digit Symbol Substitution (DSS) exercise score and the Consortium to Establish a Registry for Alzheimer's Disease (CERAD) total score. Healthy eating index (HEI), PA, and sleep problem and hours of sleep were assessed by questionnaire. The association between cognitive function and HEI, PA, sleep problem and hours of sleep were assessed by linear regression after adjusting for age, gender, race/ethnicity, poverty level, lipid profile, fasting glucose level, alcohol, body mass index, stroke and education. Data were analyzed using Stata 14 considering design and sample weight and p<0.05 is statistically significant. **RESULTS/ANTICIPATED RESULTS:** CERAD total score was associated with HEI (Adjusted B = 0.07, 95% Confidence Interval