

## Oral presentations

### OP-004 THE EPIDEMIOLOGY AND HEALTH SYSTEM IMPACT OF MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY AMONG AFFECTED CHILDREN AND THOSE WITH FALSE POSITIVE NEWBORN SCREENING RESULTS IN ONTARIO, CANADA

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**Introduction** Limited research has focused on the health system impact of rare genetic disorders, including inborn errors of metabolism (IEM). Investigating patterns of health services use and their association with social and geographic characteristics is important for understanding the burden of disease, including the impact of screening and clinical management; and for identifying potential inequities in access to care.

**Objectives** We conducted an observational study to provide a comprehensive description of the epidemiology and health system impact of IEM in Ontario, beginning with medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

**Methods** The study cohorts consist of Ontario infants diagnosed with MCADD following a positive newborn screening result from April 2006 through March 2010 (n=45); and those who received a false positive screening result for MCADD during the same time period (n=51). The primary control population includes a random sample of infants with negative screening results. Two distinct secondary control groups were created by matching to the truly affected cohort and the false-positive cohort, respectively. Screening and confirmatory testing results are securely linked at the individual level with population-based administrative databases encompassing health services use (physician visits, hospitalizations, and emergency department care) for all insured Ontarians from April 2006 through March 2012. Based on the results of a literature review we conducted to identify existing approaches to addressing statistical reliability with small sample sizes, analyses are largely descriptive.

**Results** We will describe: (i) the patterns of health care services use overall and by sociodemographic and geographic characteristics, (ii) the associations between types of service use and proxy health outcomes, and (iii) the costs of care.