4th International Conference on
PEDIATRICS AND NEONATOLOGY
November 18-19, 2024 | Bangkok, Thailand

TITLE: Galactosemia among Positive-screened Patients who Underwent Lactose Challenge: A Review of Records of the Newborn Screening Program

Name: Mary Erika V. Orteza
Affiliation: Consultant/ Center for Human Genetics Services, National Institute of Health-Manila
Country: Philippines
Email ID: mvorteza@up.edu.ph

ABSTRACT (upto 300 words)

Background. Newborns screened positive for Galactosemia through Expanded Newborn Screening (ENBS) with borderline levels undergo lactose challenge that requires interruption of breastfeeding temporarily then shifting to soy-based formula.
Objective. To determine the percentage of Classical Galactosemia (CGal), Non-classical Galactosemia (NCGal), probable mild variant form, and negative Galactosemia among newborns screened positive for Galactosemia who underwent lactose challenge.
Methods. This is a retrospective study. NBS records were reviewed, and data were collected from January 2015 to December 2020.
Results. Out of the 117 newborns screened positive for Galactosemia, 58 underwent lactose challenge. Majority were male, term with a birth weight of 2500-4000g and received a final disposition in 4-6 months. Fifteen patients underwent 1-week lactose challenge wherein six reached a resolution on first challenge. Majority, 35 (60.3%) were negative for Galactosemia, six (10.3%) probable mild variant Galactosemia, three (5.2%) NCGal, and no CGal were observed. Fourteen suspected cases (24.1%) are pending final disposition.
Conclusion. This study describes the demographics of newborns flagged for Galactosemia who underwent lactose challenge. A 1-week lactose challenge may be recommended to further detect patients who are negative for Galactosemia.

Keywords: galactosemia, ENBS, lactose challenge

BIOGRAPHY (upto 200 words)

Dr. Mary Erika V. Orteza had her Doctor of Medicine at Saint Louis University, Baguio City Philippines (2008 - 2012) and passed the Physician Licensure Examination last August 2013. She had her Internship at Saint Louis University- Hospital of the Sacred Heart and Pediatric residency Training at Baguio General Hospital and Medical Center (2014- 2017). She was a Medical Specialist II at Nueva Vizcaya Provincial Hospital, Bambang, Nueva Vizcaya (June 2017- November 2018), Medical Officer at Pines City Doctors Hospital (August 2017- November 2018) and Medical Officer IV (December 2018-2020) at Baguio General Hospital before she went to Fellowship Training on Clinical and Metabolic Genetics at Philippine General Hospital, Manila (January 1, 2020 to December 31, 2021).

Currently, she is a Medical Officer IV at Baguio General Hospital and a Geneticist consultant at Center for Human Genetics Services, National Institute of Health- Manila. She heads the Genetics Subspecialty OPD of BGH and is a member of the Newborn screening Continuity Clinic of Cordillera Administrative Region, Benguet, Philippines. She is a member of Baguio Benguet Medical Society and Philippine Medical Association (September 2017), Philippine Pediatric Society- Northern Luzon Chapter (2019) and a Diplomate of the Philippine Pediatric Society.

Presenter Name: Dr. Mary Erika V. Orteza
Mode of Presentation: Oral/Poster.
Contact number: +63 915 413 0402

Upload your photo here