Joshua Hellmann Foundation Newborn Metabolic Screening Program

Brief report
3 September 2021

Centre of Inborn Errors of Metabolism
The Chinese University of Hong Kong

With the generous support of the Joshua Hellmann Foundation, the Newborn Metabolic Screening Program has been commenced since July 2013. We would like to take this opportunity to express our sincere gratitude to all the collaborating hospitals and clinics for their effort and determination to make this meaning service available to our newborn babies.

Below are highlights of our accomplishments:

1. From July 2013 to July 2021, more than 83,585 local newborn babies were screened.
2. We were successful to keep the re-call rate below 0.350%.
3. Over 97% of the screening tests were completed within 3 calendar days.
4. Sixteen babies having different Inborn Errors of Metabolism (IEM) were identified. The diagnoses were medium-chain acyl-CoA dehydrogenase deficiency (n = 2), carnitine acylcarnitine translocase deficiency (n = 1), 2-methylbutyryl-CoA dehydrogenase deficiency (n = 1) and methionine adenosyltransferase deficiency (n = 1), neonatal intrahepatic cholestasis caused by citrin deficiency (n = 2), hyperphenylalaninaemia (n = 2), Beta-ketothiolase deficiency (n =1), primary carnitine uptake defect (n =2), Holocarboxylase synthetase deficiency (n =1), 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency (n =1), glutaric acidemia type II (n =1), congenital adrenal hyperplasia (n =1).
   All patients have been referred and managed at the Joint Metabolic Clinic of Prince of Wales Hospital.
5. One maternal condition (maternal carnitine uptake defect) (n = 4) was identified through follow up of a newborn with very low free carnitine level.
6. The overall incidence of IEM is 1 in 4,399.
7. Screening for cystic fibrosis was added to the program in Mar 2014.
8. Screening for congenital adrenal hyperplasia (CAH) was added to the program in Feb 2016.
9. Screening for X-linked adrenoleukodystrophy (X-ALD), spinal muscular atrophy (SMA) and severe combined immunodeficiency (SCID) were added to the program in Sep 2021.

Last but not the least, we would like to extend our sincerest thanks to Joshua Hellmann Foundation for the tremendous support.