



JOSHUA HELLMANN  
FOUNDATION  
FOR ORPHAN DISEASE

Joshua Hellmann Foundation Newborn  
Metabolic Screening Program

**Brief report**  
**11 October 2019**

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 started 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 June 2019, more than 63,095 local newborn babies were screened.
2. We were successful to keep the re-call rate below 0.268%
3. Over 97% of the screening tests were completed within 3 calendar days.
4. Twelve 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 = 1), hyperphenylalaninaemia (n = 2), Beta-ketothiolase deficiency (n = 1), primary carnitine uptake defect (n = 1), Holocarboxylase synthetase deficiency (n = 1), 6-pyruvoyl-tetrahydropterin synthase (PTPS) deficiency (n = 1).  
All patients have been referred and managed at the Newborn screening Clinic of Prince of Wales Hospital.
5. Four maternal conditions (maternal carnitine uptake defect) (n = 4) were identified through follow up of a newborn. The patients were referred to adult metabolic team for management at Princess Margaret Hospital. .
6. The overall prevalence of IEM is 1 in 3,943.
7. Screening for cystic fibrosis was added to the program in Mar 2014. There is no screened positive case so far.
8. Screening for congenital adrenal hyperplasia (CAH) was added to the program in Feb 2016. There is no screened positive case so far.

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