

Supplementary material

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Screening procedure	Conditions in the NBSIEM	
1. The NBSIEM is offered by the Hospital Authority.	1. Beta-ketothiolase deficiency	
2. All Hong Kong Chinese parents are offered newborn	2. Glutaric acidaemia type I	
screening for their children born at public hospitals.	3. Isovaleric acidaemia	
Approximately 50 000 Chinese newborns are	4. Methylmalonic acidaemia (methylmalonyl-coenzyme A mutase	
delivered at public hospitals each year. Of these,	deficiency)	
around three-fifths will benefit from this	5. Methylmalonic acidaemia and homocystinaemia (cobalamin C	
programme. ²	deficiency)	
3. A few drops of blood are obtained from a heel prick	6. Multiple carboxylase deficiency	
and collected on a filter paper card between 24 to 72	7. Propionic acidaemia	
hours after birth. The card with dried bloodspots is	8. 3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency	
sent to a screening laboratory and subjected to	9. Argininaemia	
biomarker analysis.	10. Argininosuccinic acidaemia	
4. Under the current policy, the card is discarded after	11. Citrullinaemia type I	
completion of quality assurance.	12. Citrullinaemia type II	
	13. Classic phenylketonuria	
	14. Homocystinuria	
	15. Maple syrup urine disease	
	16. Tyrosinemia type I	
	17. 6-pyruvoyl-tetrahydropterin synthase deficiency	
	18. Carnitine-acylcarnitine translocase deficiency	
	19. Carnitine palmitoyltransferase II deficiency	
	20. Carnitine uptake deficiency	

Supplementary Table 1. Summary of the Newborn Screening Programme for Inborn Errors of Metabolism (NBSIEM) in Hong Kong¹

21. Glutaric acidaemia type II
22. Medium-chain acyl-coenzyme A dehydrogenase deficiency
23. Very long-chain acyl-coenzyme A dehydrogenase deficiency
24. Biotinidase deficiency
25. Classic galactosaemia
26. Congenital adrenal hyperplasia
27. Severe combined immunodeficiency

References

- Hospital Authority. Newborn Screening Programme for Inborn Errors of Metabolism (IEM). October 2023. Available from: <u>https://www21.ha.org.hk/smartpatient/SPW/MediaLibraries/SPW/SPWMedia/Education-pamphlet_NBS-IEM_Eng-(Oct-2023).pdf?ext=.pdf</u>. Accessed 27 Mar 2024.
- 2. Census and Statistics Department, Hong Kong SAR Government. Hong Kong Annual Digest of Statistics (2021 Edition). October 2021. Available from: https://www.censtatd.gov.hk/en/data/stat_report/product/B1010003/att/B10100032021AN21B0100.pdf. Accessed 1 Sep 2022.

	No. (%)
Profession	
Doctor	15 (50.0%)
Nurse/midwife	13 (43.3%)
Academic	1 (3.3%)
Phlebotomist	1 (3.3%)
Specialty	
Obstetrics and gynaecology	13 (43.3%)
Paediatrics	13 (43.3%)
Pathology/laboratory	4 (13.3%)
Sector	
Public	28 (93.3%)
Private	2 (6.7%)
Years of experience	
<5	4 (13.3%)
5-10	12 (40.0%)
>10	14 (46.7%)

Supplementary Table 2. Interviewee characteristics (n=30)