This article was published on 16 Jul 2024 at www.hkmj.org.

Secondary use of dried blood spots from newborn screening

Hong Kong Med J 2024;30:338 https://doi.org/10.12809/hkmj2411850

To the Editor—The opt-in, territory-wide Newborn Screening Programme for Inborn Errors of Metabolism relies on collection of a newborn's dried blood spot (DBS) at birth. The residual DBS (rDBS) samples contain genetic material and may be stored for secondary research purposes. Nonetheless according to the findings by Ngan et al,¹ not all healthcare professionals are confident in explaining to hesitant parents the secondary benefits of the Programme. Below is an example of how rDBS may be utilised for translational research through analysis of genes in samples.

lymphoblastic Acute leukaemia is а common childhood malignancy of multifactorial pathogenesis and may present before 12 months of age²; such an early onset has inspired research into any prenatal gene abnormalities in diagnosed individuals. Related chromosomal aberrations such as *BCR-ABL1* translocation and high hyperdiploidy have been detected in rDBS samples of individuals subsequently diagnosed with acute lymphoblastic leukaemia,³ suggesting in-utero involvement. Closer monitoring of genetically high-risk children may facilitate timely detection of any cancer, although such cost-effectiveness is yet to be assessed.

Clinical application of rDBS research is still in its infancy but has potential in populationwide disease tracking and epidemiological studies. Equipping healthcare professionals with updates on relevant studies may facilitate communication with prospective parents. It is hoped that more parents will opt in and contribute precious rDBS samples to the scientific community.

Author contributions

Concept or design: CWM Leung. Acquisition of data: CWM Leung. Analysis or interpretation of data: CWM Leung. Drafting of the manuscript: CWM Leung, NS Cheng. Critical revision of the manuscript for important intellectual content: NS Cheng, TF Leung. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of interest

All authors have disclosed no conflicts of interest.

Acknowledgement

The authors thank Mr Samson Mak for providing medical editing support.

Funding/support

This letter received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Christy WM Leung¹, MB, ChB NS Cheng², DNurs TF Leung^{2,3}*, MD, FRCPCH

- ¹ Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong SAR, China
- ² Department of Paediatrics, The Chinese University of Hong Kong, Prince of Wales Hospital, Hong Kong SAR, China
- ³ Hong Kong Hub of Paediatric Excellence, The Chinese University of Hong Kong, Hong Kong SAR, China

* Corresponding author: tfleung@cuhk.edu.hk

References

- Ngan OM, Tam CJ, Li CK. Exploration of clinical and ethical issues in an expanded newborn metabolic screening programme: a qualitative interview study of healthcare professionals in Hong Kong. Hong Kong Med J 2024;30:120-9.
- Cheng FW, Lam GK, Cheuk DK, et al. Overview of treatment of childhood acute lymphoblastic leukaemia in Hong Kong. Hong Kong J Paediatr (new series) 2019;24:184-91.
- Rüchel N, Jepsen VH, Hein D, Fischer U, Borkhardt A, Gössling KL. In utero development and immunosurveillance of B cell acute lymphoblastic leukemia. Curr Treat Options Oncol 2022;23:543-61.