

Views towards dried blood spot cards storage and its usage for extended genetic testing among parents and healthcare providers in Hong Kong

(Parents)

The survey aims to gather attitude towards dried blood spot card storage and extended genetic testing among parents, informing public policy development in future. The study is funded by the Health and Medical Research Fund, the Food and Health Bureau and conducted by the Hong Kong Children's Hospital, in collaboration with Pamela Youde Nethersole Eastern Hospital, Princess Margaret Hospital, and the Faculty of Medicine of the University of Hong Kong.

The survey is anonymous, and your participation is entirely voluntary. All information that you provide is kept secure. It approximately takes about 10-15 minutes. The study has been reviewed by the Hong Kong Children's Hospital Research Ethics Committee, Kowloon West Cluster Research Ethics Committee, and Hong Kong East Cluster Research Ethics Committee.

** Information provided would not affect the care your child receives.

Part IA Basic Information

1. Your age: _____ years old
2. Gender: Male Female
3. What is your ethnicity?
 Chinese (Hong Kong) Chinese (Mainland China) Other, please specify _____
4. Are you a Hong Kong permanent resident?
 Yes No, How long have you been living in Hong Kong? _____ years
5. What is your religious affiliation?
 None, I have no religious preference
 Christian
 Catholic
 Buddhist
 Other, please specify _____
6. Are you and your partner consanguineous?
 No Yes Not Sure
7. Do you work in a health-care related field?
 No Yes
8. What is your highest level of education achieved?
 Primary school Lower secondary school (Secondary 1-3)
 Upper secondary school (Secondary 4-7) Post-secondary (diploma/ associate degree)
 Tertiary / Bachelor (University) Master's degree or above
9. What is your family household monthly income (HKD)?
 Below \$10,000 \$10,000-\$19,999 \$20,000-\$29,999
 \$30,000-\$39,999 \$40,000-\$49,999 \$50,000 or above

Part IB Basic Information- About your child(ren)

1. How many children do you have?
 1 2 3 4 or more
2. Has/have your child/children been diagnosed with any medical diseases / conditions?
 No Yes, please specify _____

Description of Newborn Screening with Dried Blood Spot Card (DBS card)

Shortly after your baby is born, a health professional will perform a heel prick to collect a few drops of blood on a special card. The DBS card will be sent to the laboratory for the screening of some rare but serious conditions which could be picked up and treated early with benefit the affected babies in the long run. The DBS card also increasingly secondary uses, such as diagnosis of infective conditions, patient specific treatment, scientific research and extended genetic testing.

DBS Cards are stored by various countries for various duration ranging from 6 months to indefinitely after Newborn Screening has been completed for secondary uses i.e. quality assurance.

1. Have you heard about Newborn Screening for uncommon disorders in Hong Kong?

No → Please go to Q. 2

Yes → Please go to Q. 1.1

1.1 From which of the following sources have you heard of Newborn Screening? (Check all that apply)

Hospital websites or brochure

Healthcare provider, including doctors and nurses

Research papers

Other parents

Media (i.e., newspaper, magazine)

Websites, online platforms

Others, please specify: _____

1.2 Has your child / have your children taken part in Newborn Screening for uncommon disorders?

No

Yes

Not Sure

1.3 From your experience, at which time point did you receive the information about the Newborn Screening?

First Trimester Pregnancy (1-12 weeks)

Second Trimester Pregnancy (13-26 weeks)

Third Trimester Pregnancy (27-40 weeks)

Soon after the birth delivery (First or second day)

Not Sure

1.4 Did you think Newborn Screening was useful?

No

Yes

2. **This section asks about your understanding of the DBS card. Please decide whether the following sentences are true or false to the best of your knowledge.** (Please tick appropriate box)

a) The DBS card can identify rare and serious conditions in babies	<input type="checkbox"/> True	<input type="checkbox"/> False
b) The DBS card can be stored for a long period of time	<input type="checkbox"/> True	<input type="checkbox"/> False
c) There is genetic material (DNA) in the DBS card	<input type="checkbox"/> True	<input type="checkbox"/> False
d) If DBS cards were to be stored, parents' consent is required	<input type="checkbox"/> True	<input type="checkbox"/> False
e) The DBS card can support scientific research	<input type="checkbox"/> True	<input type="checkbox"/> False
f) The DBS card can aid public health research and future policy implementation	<input type="checkbox"/> True	<input type="checkbox"/> False
g) The DBS card can be used for quality assurance and development of new screening tests	<input type="checkbox"/> True	<input type="checkbox"/> False
h) The DBS card can aid disease treatment or be used for the development of individual specific disease treatment	<input type="checkbox"/> True	<input type="checkbox"/> False
i) The DBS card can be used for diagnostic purpose in the future (i.e. neonatal infection with presentation in infancy / childhood)	<input type="checkbox"/> True	<input type="checkbox"/> False
j) The DBS card can be used for forensic purposes	<input type="checkbox"/> True	<input type="checkbox"/> False
k) The DBS card can be connected to personal medical record	<input type="checkbox"/> True	<input type="checkbox"/> False

3. **The following lists out concerns related to the storage of DBS card.** Please use the following scale to indicate how much you concern with each statement below from 1= Not at all to 5= Extremely concerned. (Please tick appropriate circle)

Statements	Not Concerned			Extremely	
	At All	-----	-----	Concerned	
a) The DBS card is stored for public health and scientific research. Would you be concerned?	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5
b) The DBS card is shared with research teams among academic institutions. Would you be concerned?	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5
c) The DBS card will be linked with personal or medical information. That is, it is not stored anonymously. Would you be concerned?	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5
d) The subsequent research does not provide direct benefit towards my baby / child. Would you be concerned?	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5
e) Would you be concerned if the DBS card would lead to breaches of personal data?	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5

4. How long do you think residual DBS card should be stored?

- Up to 6 months (e.g. current HA policy)
- Up to 2 years (e.g. Texas, U.S.)
- Up to 5 years (e.g. Netherland)
- Up to 18 years (e.g. NSW, Australia)
- Indefinitely (e.g. Denmark)

5. Overall, would you support the DBS card storage? (Please circle the score that best fits your feelings)



Not support at all

Very supportive

Part IIB Understanding of Extended Genetic Testing

Description of Extended Genetic Testing

It is possible to study a person's entire genome extracted from DBS card. This process is called extended genetic sequencing. It may give you information about a person's risk of having different diseases both in the present and in the future, including childhood diseases such as epilepsy and adult onset diseases breast cancer or diabetes.

Imagine that you have a recently born baby. Newborn Screening program now offers you the chance to get extended genetic sequencing done for your baby. You would receive the results and would not have to pay for the testing. You can decide whether or not you want the information to be a part of your child's medical record.

1. Have you heard of extended genetic testing in Newborn Screening?

- No
- Yes

2. Below are some scenarios describing the return of extended genetic testing results. If you were the parent, would you wish to receive the following information with regards to your child? (Please tick appropriate box)

a) A condition list on the current medical recommended screening panel	<input type="checkbox"/> No	<input type="checkbox"/> Yes
b) The newborn is a carrier for the condition but will not develop it	<input type="checkbox"/> No	<input type="checkbox"/> Yes
c) A childhood-onset disorder is a condition with treatment i.e. epilepsy, Maturity-onset diabetes of the young(MODY)	<input type="checkbox"/> No	<input type="checkbox"/> Yes
d) A childhood onset disorder is a condition with NO available treatment i.e. Lowe syndrome	<input type="checkbox"/> No	<input type="checkbox"/> Yes
e) An adulthood-onset disorder is a condition with treatment i.e. hereditary breast and ovarian cancer	<input type="checkbox"/> No	<input type="checkbox"/> Yes
f) An adulthood onset disorder is a condition with NO available treatment i.e. Alzheimer's disease and Huntington's disease	<input type="checkbox"/> No	<input type="checkbox"/> Yes

g) Increase risk for an adult-onset disease i.e. heart disease, diabetes	<input type="checkbox"/> No	<input type="checkbox"/> Yes
h) A variant that has unknown clinical implications	<input type="checkbox"/> No	<input type="checkbox"/> Yes

3. What is the factor(s) influencing your interest in extended genetic testing? Please use the following scale to indicate how important each factor would be in your decision to have testing from 1 = Not at all to 5 = Very important. (Please circle appropriate number)

Factors	Not Important ----- Important				
	At All	1	2	3	4
a) Accuracy of the test results/ sequencing results	1	2	3	4	5
b) Diagnosing susceptibility to adult onset disease (i.e. breast cancer / colon cancer / diabetes) during newborn period	1	2	3	4	5
c) Access to specialist follow-up for affected individuals	1	2	3	4	5
d) Access to existing treatment for affected individuals	1	2	3	4	5
e) Shorten the diagnosis time in future	1	2	3	4	5
f) Identify new disease genes and diagnose individuals with rare disorders	1	2	3	4	5
g) Impinge on the child's right to an open future	1	2	3	4	5

4. How interested would you be in getting your child's extended genetic testing?



Not interested at all

Very interested

Thank you for your time and interest in completing this questionnaire!