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Position Statement

Title:	**Supplemental, Online (Direct to Consumer) DNA Newborn Screening Tests**
Document Number:	2019PS02
Publication Date:	October 2019
Replaces:	2018PS03
Review Date:	October 2022

Background
Recently, online, Direct-to-consumer (DTC) genetic tests designed to be used in newborns and young children have begun to be offered in the Australasian market. These commercial tests typically involve an at-home cheek-brush DNA collection which is sent to a laboratory to test for a panel of genetic disorders. The tests may be advertised as being designed to be used in conjunction with conventional Government funded newborn bloodspot screening (Gov funded NBS) testing.

Gov funded NBS is a well-established and carefully planned public health program which is funded and supported by government health departments in Australia and New Zealand. It is available for all newborns at no cost to parents. The disorders on NBS panels have been carefully selected by medical and scientific experts to ensure the tests are as effective as possible with the least possible harm. There are well-established pathways for the follow-up and treatment of babies with abnormal results within the public health system. The same will be true for new tests which will be added over the coming years.

Parents have to pay for DTC genetic testing. While the decision to perform one of these DTC genetic tests is at the discretion of parents, it is important that new parents are not confused by this type of testing and that they continue to have Gov funded NBS performed on their babies. In particular:

1. DTC genetic testing is NOT a substitute for Gov funded NBS. For example, congenital hypothyroidism, which occurs in ~1:2000 babies, is the most common disorder detected by Gov funded NBS. DTC genetic testing will NOT detect most cases of congenital hypothyroidism.

2. Parents should continue to be informed about and encouraged to have Gov funded NBS performed on their babies at 2 to 3 days of age, regardless of any other tests that may be planned.

3. In making the decision whether or not to have an online DTC genetic test on their

newborn, parents should carefully consider

- The range of conditions offered, in particular:
    - Is there treatment available (and funded) which is effective before symptoms develop?
    - Will there be difficulty deciding if the genetic test truly indicates that a disorder will certainly develop?
    - Does the 'disorder' need treatment?
    - Will the result be available in time to ensure effective treatment (some conditions present within days of birth and must be treated at that time)?
    - Does the condition present in later life and if so, would you want to know about it?
- Who will get the results and be able to interpret them?
- Where to get more information about the testing and the disorders (is pre- and post-test counselling available)?

This policy is in line with recent recommendations for example 'Health care professionals should recommend against use of direct-to-consumer newborn sequencing services by parents.' *Sequencing Newborns: A Call for Nuanced Use of Genomic Technologies.* Johnson J, Lantos JD, Goldenbery A, Chen F, Parens E, Loenig BA and members of the NSIGHT Ethics and Policy Advisory Board. First published: 14 August 2018 https://doi.org/10.1002/hast.874

A further HGSA statement outlining the issues of online DNA testing can be found at: https://www.hgsa.org.au/documents/item/18