

Monday, 4 March 2013

Dr Jane Hendtlass  
Coroner  
Coroners Court of Victoria  
Level 11, 222 Exhibition Street  
Melbourne Vic 3000  
Via email



Dear Coroner Hendtlass,

**Subject: Court Reference COR 2006 001014**

I refer to your recommendations:

Recommendation 2. That Victorian Clinical Genetics Services establish genetic screening for babies who would otherwise undergo liver biopsies and have characteristics associated with increased likelihood of citrin deficiency

And

Recommendation 3. That Victorian Clinical Genetics Services provide genetic screening for parents of children who have been identified as carriers of genetic screening.

An alternative to the coroner's recommendation has been implemented.

Since the death of Alyssa Chan in 2006 there has been significant improvement in the availability of genetic testing for citrin deficiency. There are now at least seven laboratories around the world offering this test as a routine clinical service, with a turnaround time of between three and five weeks. VCGS currently offers a state wide clinical genetics service that upon request, is able to assist with sample collection, DNA extraction and shipping to facilitate genetic testing for citrin deficiency. Funding for this type of testing needs to be provided by the referring hospital.

It is worth noting that there are now approximately 3000 different diagnostic genetic tests available and it is not practical for any single laboratory to perform any more than a small fraction of these tests. Given that genetic testing for citrin deficiency is now readily available and with rapid turnaround times, it is neither cost effective nor clinically necessary for VCGS to establish this as an 'in house' test.

Should you need further information, please do not hesitate to contact me.

Yours sincerely,

A handwritten signature in black ink, appearing to read "David Amor".

Associate Professor David Amor

Director, Victorian Clinical Genetics Services