BC CHILDREN'S HOSPITAL
An agency of the Provincial Health Services Authority

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Women's
BC WOMEN'S HOSPITAL & HEALTH CENTRE
An agency of the Provincial Health Services Authority

Jul 14, 2014

Lisa Lapointe
Chief Coroner of BC
Office of the Chief Coroner
Metrotower II
Suite 800 4720 Kingsway
Burnaby BC V5H 4N2

Dear Lisa

Dr Tanya Nelson forwarded your letter and the "BC Coroners Service Child Death review panel Report and Recommendations" to me and asked that I respond specifically to you regarding the following recommendation in relation to the CPT I variant:

Recommendation 2: Genetic Testing

□ That criteria be established by the BC Coroners Service, in consultation with pathologists and clinical geneticists, and the First Nations Health Authority, to identify when genetic testing would be beneficial in helping to establish a cause of death in infants.

□ The BC Coroners Service, pediatric pathologists, the Provincial Medical Genetics Program and the First Nations Health Authority review the utility of testing for the CPT1a variant in First Nations infants.

For the past 2 years, a multidisciplinary working group of health care providers, facilitated by the Ministry of Health, have been working on a strategy to mitigate any health risks associated with the CPT1 variant. We have developed a Medical Guideline and Parent brochure which are near completion. Recently, the Ministry of Health has transferred coordination of this effort to the First Nations Health Authority. The Parent brochure is focused on prevention of hypoglycemia through the education of parents on healthy feeding practices, avoidance of prolonged fasting and signs of illness that should prompt a parent to seek medical attention.
Likewise, the Medical Guideline provides evidence-based information on the prevention and management of hypoglycaemia including the utility of testing for the CPT Ia variant. The key message is that clinicians need to provide information to First Nations' families about healthy feeding practices with an aim to prevent hypoglycaemia, especially during intercurrent illness or during periods of prolonged fasting. Health care providers are encouraged to integrate this information with other key prevention and health promotion strategies including breastfeeding and safe sleep practices.

The criteria for when to request CPT Ia genetic testing in the setting of sudden unexpected death was not within scope of the Medical Guideline. However, the current practice has been to request review of the original newborn screening acylcarnitine profile. Approximately 95% of infants with the CPT I variant have an elevated ratio of free carnitine (C0) to long chain acylcarnitine species (elevated C0/ C16+C18). CPT I molecular testing is available in the Molecular Genetics laboratory, C&W, for confirmation.

I would be happy to participate in a meeting to review the current practice and reach a consensus on when CPTIa genetic testing would be appropriate in the investigation of FN infant death. Feel free to contact me to discuss further.

Sincerely,

Vallance

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cc: Dr Tanya Nelson