Dr Deepak Gill, MB BS FRACP
Paediatric Neurologist
T. Y. Nelson Department of Neurology and Neurosurgery
Phone: +61 2 9845 2694 Fax: +61 2 9845 3905
Provider No. 2327163T

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The Australian Senate Community Affairs Committee
PO Box 6100
Parliament House
Canberra ACT 2600

Submission to the Senate Community Affairs Committee Inquiry into Gene Patents

I thank you for the invitation to attend the panel session on 5 August 2009. I am unable to attend the hearing, however I would like to submit the current summary of the availability of testing of SCN1A.

Genetic investigations are an increasingly important aspect in the diagnosis in childhood neurological disorders. In early childhood onset epilepsy mutations in the gene encoding for the alpha subunit of the neuronal sodium channel (SCN1A) cause a severe form of epilepsy, severe myoclonic epilepsy of infancy, also known as Dravet syndrome. The prevalence of Dravet syndrome is estimated at 1:40,000 of the population, but may be more common than this. The SCN1A gene has been recognised as the most important gene discovery, and clinically important gene, in childhood epilepsy.

Early and prompt diagnosis in Dravet syndrome leads to treatment with appropriate medications and reduces the risk of recurrent seizures and recurrent status epilepticus. The diagnosis of Dravet syndrome may be difficult to confirm early in the course of the condition on clinical grounds alone, and the epilepsy of Dravet syndrome may mimic other epilepsies of infancy. This further states the clinical need for the availability and access to testing for all children with possible new onset Dravet syndrome.

Currently the testing for SCN1A in Australia is restricted to a single commercial laboratory who holds the license and rights to the testing for this gene. Although the company states that that they " have never sought to refuse to license others in areas covered by our patent portfolio. Indeed we have a corporate mission of actively seeking to engage with others in order to facilitate broad licensing of our rights." This has yet to be demonstrated and tested. This engagement needs to be completed and an outcome that allows clinical testing and ready access to the results with the relevant suitable expert interpretation will be the way to demonstrate that there is going to be no adverse impact on the health and wellbeing of Australian patients with early onset epilepsy and possible Dravet syndrome.

The support for research is commended and crucial and a good example of this is the field epilepsy genetics. A laboratory that performs the testing should be part of a research program aiming to define the phenotype/genotype correlations, or the results should be available to allow others to do this.

I am not aware of any evidence that the licensing of one laboratory to carry out the tests has had a significant negative impact on research in the field of SCN1A testing, however more data available from a more widely available test could provide more research opportunities to study the evolution and mechanism of disease causing genes and epilepsy.

With the current testing of SCN1A, samples from our public hospital when sent overseas, may take up to 3 for the result to be available. The result is valued in that it includes a detailed interpretation from a paediatric neurologist, and that the result is part of an on-going research program aiming to find the mechanisms of SCN1A and epilepsy. It is this interaction with clinician rather than a mechanistic approach that adds value to a test, and this value should be considered when assessing the usefulness and cost-effectiveness of genetic testing. Any patent or license should allow this to happen within Australia.

If you would like any further information on this matter please do not hesitate to contact me.

Kind regards

Yours sincerely

Deepak Gill
Paediatric Neurologist