

**Question on Notice
No. 661
Asked on 9 May 2006**

MS MALE asked the Minister for Health (MR ROBERTSON)—

QUESTION:

Will he advise of the additional newborn screening processes that are available at Queensland Health through the purchase and utilisation of the Tandem Mass Spectrometer, and what additional outcomes and diagnoses have been made?

ANSWER:

The Tandem Mass Spectrometer was purchased and installed in March 2003. It was fully commissioned and validated for use in late 2003. It has been running at full capacity since April 2004.

Having this instrument means that several rare but serious diseases can now be diagnosed more rapidly through analysis in Queensland. These include the Medium Chain Acyl CoA Dehydrogenase Deficiencies (also known as MCAD's) and diseases causing irreversible neurological damage associated with Vit B12 deficiencies.

Having this instrument also means more rapid diagnoses of the various metabolic causes of severe neonatal hypoglycaemia and assisting in the diagnoses of organic acid metabolic disorders. It also allows timely follow up in monitoring the patient's response to treatment.

All these diseases need to be detected early and treatment begun as soon as possible to halt the progression of deleterious effects of the diseases. Delays in diagnosis mean less favourable outcomes are likely. If this testing was not done and the diseases went undetected, 30-40% of patients affected would die very young and another 30-40% would have severe mental retardation.

Between April 2004 and July 2005 67,000 Queensland and 3,800 Interstate and Overseas babies have used this service. Of these 70,800 babies 31 have been detected with serious life shortening and neurologically degenerative inborn errors of metabolism. There were eight MCAD's, 10 Vit B12 Neurological Diseases and 13 other diseases. Previously all these would have been sent interstate for diagnosis with the necessary additional delays in reaching a diagnosis for the patients due to transportation and capacity factors in interstate Labs.

In part because of the earlier diagnosis with this instrument in Queensland allowing treatment to start earlier all 31 patients are alive and are doing well.