No increase in commonest preventable cause of intellectual disability over 20 years

July 19 2011

A new study that was prompted by recent reports of an increase in cases of congenital hypothyroidism in the United States, and aimed at assessing the incidence of this condition among Quebec newborns, suggests that the increase is entirely artifactual.

CH is characterized by inadequate thyroid hormone production and is the most common cause of preventable intellectual disability. The results were published in the Journal of Clinical Endocrinology and Metabolism by Dr. Johnny Deladoëy, a pediatric endocrinologist and researcher in metabolic and genetic diseases at CHU Sainte-Justine Research Center and an assistant professor of pediatrics and biochemistry at University of Montreal.

The increase in the number of cases identified may be an artifact due to changes in screening methods and results in the treatment of some children who may, in fact, not benefit from treatment. "Overly sensitive screening methods that identify mild cases of CH represent an obvious shift in neonatal screening from its original purpose, which was to identify severe cases in which the benefits from treatment would be clearly documented", Deladoëy said. "This might explain why, in the United States, more than a third of children labelled as having CH on the basis of neonatal screening no longer receive treatment after four years of age."

The lack of consensus about the thresholds on which to base detection has always presented a challenge for CH screening. Quebec pioneered

universal screening for CH in newborns, being the first jurisdiction on the globe to initiate such a program in 1973. Quebec strategies and methodologies have proven to deliver reliable results and may serve as a model in the standardization of CH screening and diagnosis worldwide.

Thanks to a contribution rate of more than 90 % of hospitals and physicians to a provincial database over a period of 20 years, the results from the detection and diagnosis of 620 cases of CH could be analyzed.

Details about the study:

Historical data were analyzed for the study jointly with the Québec newborn blood screening laboratory located at the Centre hospitalier universitaire de Québec (CHUQ). More precisely, of the 1,660,857 newborns tested over the 20-year period, 620 cases of CH were detected, diagnosed and treated. During the second half of the period, a slightly more sensitive detection threshold was applied – in addition to those expected, 49 further cases of CH were detected, representing an 18 % increase. Most of these additional cases had mild functional disorders whose impact on mental development is uncertain. Most importantly, the number of detected cases would have remained stable throughout the period studied if the screening threshold had been unchanged.

The study was published ahead of print on June 1, 2011 in the Journal of Clinical Endocrinology and Metabolism by Dr. Johnny Deladoey of the CHU Sainte-Justine Research Center and University of Montreal, jointly with Dr. Guy Van Vliet, also of the CHU Ste-Justine, and Dr. Jean Ruel and Dr. Yves Giguère, of the Québec newborn blood screening laboratory at the CHUQ.

Provided by University of Montreal