

New gene test discovered for inherited neuromuscular disorder

Newcastle University scientists have identified a new gene which will allow rapid diagnosis and earlier treatment of a debilitating neuromuscular condition.

The gene, GFPT1, is crucial in causing a variation of Congenital Myasthenic Syndrome (CMS) which gained media attention recently with the plight of baby RB, who was at the centre of a “right-to-life” legal dispute.   
  
CMS is a rare genetic condition affecting the way signals travel between the brain and muscles which can cause paralysis and in some cases death. It affects one in every 500,000 births and the severity of the condition varies, depending on where the fault lies in the complex signals between the nerves and the muscles.  
  
The variation of CMS identified by the team of international researchers, GFPT1, tends to develop in the first ten years of life with patients losing muscle strength and control in their hips and shoulders or arms and legs.  
  
“The identification of this gene means that doctors can order genetic analysis and confirm the condition allowing earlier treatment with cholinesterase inhibitors,” explained [Professor Hanns Lochmüller](http://www.ncl.ac.uk/ihg/staff/profile/hanns.lochmuller) of the Institute of Human Genetics at Newcastle University.   
  
“This offers an effective therapy which can be taken through life,” he added.   
  
The research also highlighted a new area to explore for future treatments as GFPT1 is involved in initiating the metabolism of amino sugar.   
  
The international team, headed up by Dr. Jan Senderek from the University of Aachen in Germany and by Dr Juliane Müller from Newcastle University, analysed the genes of 13 families affected by the condition.

**Academic paper:** [Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect](http://www.sciencedirect.com/science?_ob=ArticleURL&_udi=B8JDD-524WS13-5&_user=10&_coverDate=02%2F11%2F2011&_rdoc=6&_fmt=high&_orig=browse&_origin=browse&_zone=rslt_list_item&_srch=doc-info%28%23toc%2343612%232011%23999119997%232902744%23FLA%23display%23Volume%29&_cdi=43612&_sort=d&_docanchor=&_ct=15&_acct=C000050221&_version=1&_urlVersion=0&_userid=10&md5=3e92c098d0b89c64699a8e8a0b24cf4c&searchtype=a)  
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