



Genomics England whole genome sequencing – an opportunity for rare disorders

Genomics England is a project set up by the Department of Health that is due to start in January 2014. The goal is to identify the genetic cause for all rare genetically undefined disorders and to sequence patients and families genomes to help with diagnosis and the development of future treatments.

This is a very important opportunity for patients with neuromuscular disorders to have their genome sequenced in order to maximise their chance of a genetic diagnosis. This will be important for future treatments. Weblink <http://www.genomicsengland.co.uk/>

What to do?

To have your genome sequenced we need a fresh blood sample from patients where their neuromuscular gene is not known. We also need fresh blood from their parents or at least two other close family members (siblings), with or without the problem. At the National Hospital for Neurology we are recruiting patients to be part of the first wave of sequencing and we need blood to be taken in December or January. This will give the earliest results but we will continue to collect blood after this for sequencing to include as many patients as possible in the long run.

If you wish to be part of the genome sequencing could you contact the MRC Centre for Neuromuscular Diseases in London (karen.suetterlin@ucl.ac.uk ; f.jaffer@ucl.ac.uk; g.gang@ucl.ac.uk) or write to us by post using the address below with you name, address, telephone number and which family members (we need three or more per family) could attend.

We will then arrange a date for you to come up to London either on a weekend or a week day to have the fresh blood taken. In some instances we can ask your GP to take blood to send to us.

Thank you for your help,

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