Dear colleagues,

The 100,000 Genomes Project is an opportunity for you to get a genetic diagnosis for your patients with rare disorders – FOR FREE- by Whole Genome Sequencing – please make full use of it!

"This technology has the potential to change medicine for ever but we need NHS staff, patients and the public to recognise and embrace its huge potential"

Dame Sally Davies, Chief Medical Officer

How to refer:

1. **Check patient eligibility** www.genomicsengland.co.uk/information-for-gmc-staff/rare-disease-documents/rare-disease-eligibility-criteria/
2. **Discuss with patient—**visit www.genomicsengland.co.uk for additional leaflets, videos and information
   - *the purpose of the project is to obtain a genetic diagnosis for patients with rare disorders*
   - *blood samples will be needed from affected individual(s) and unaffected parents, or from multiple affected family members*
   - *data will be made available in anonymized form to research/commercial bodies*
   - *the patient will have the opportunity (choice) to learn about 'secondary findings' – important genetic information unrelated to the primary diagnosis*
3. **Inform 100,000 Genomes Project Recruitment team rde-tr.swgmc@nhs.net**
   - Name and DOB of Proband
   - Name and DOB of parents and other affected family members *defined in criteria*
   - Level 4 disease category *defined in criteria*

What happens next:

1. Appointments, Consent and Sample handling are managed but the project team
2. You will be required to provide phenotype data (support provided)
3. Results will be returned in 12 to 18 months

Thank you

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