

Dear Colleagues

Re: Specialist Genomic Testing for Rare Diseases across South East England

We wish to inform you that from the 1st February 2021 all genomic tests should be ordered in line with the National Genomic Test Directory for rare inherited diseases (<https://www.england.nhs.uk/publication/national-genomic-test-directories/>).

In addition to the Core repertoire of Rare Disease genomic tests, you will now be able to request through the South East GLH, specialist genomic tests as defined in the National Genomic Test Directory.

Genomics tests are now commissioned and funded through NHS England and Improvement, so you will not be invoiced for tests that meet the criteria specified in the National Genomic Test Directory for rare inherited diseases. Patient eligibility criteria and guidance for appropriate testing can also be found on the national test directory web page.

The South East GLH:

NHS England have reconfigured genetic laboratory services and recently commissioned 7 national Genomic Laboratory Hubs (GLH). The South East GLH, a network of leading foundation trusts and pathology providers, is your local Genomics Laboratory Hub and has been commissioned to deliver all genomic testing services across South London, Kent, Surrey and Sussex. More information about the NHSE South East Genomic Medicine Service Alliance (GMSA) and GLH can be found here: <https://southeastgenomics.nhs.uk/>.

Requesting a Test and Sample Requirements:

This new service model may result in a change to how you order tests or to the design of the reports you receive. All South East GLH referral forms can be found on our website which also has important additional information on how to order these tests, including using the National Genomic Test Directory Clinical Indication ID, sample requirements and turnaround times (<https://southeastgenomics.nhs.uk/professionals/#how-to-order-a-test>).

Please be aware that whilst you will order a test through the South East GLH, for some specialist tests, we will send samples onto the appropriate national GLH provider and you will receive the reports directly from the testing GLH. The full list of specialist test categories are listed in Annex 1.

We would be grateful if you could cascade this information to all relevant clinical and laboratory teams within your organisation. We look forward to working with you and if you have any queries please email gst-tr.southeastGLH@nhs.net.

Yours sincerely



Richard Hall
SE GLH Ops Director



Sean Whittaker
SE GLH Medical Director



Deborah Morris-Rosendahl
SE GLH Scientific Director

Annex 1: Genomic Specialist Test Categories

RD Specialist Service	Main Providers of Speciality
Cardiology	South East GLH
Gastrohepatology	South East GLH
Haematology	South East GLH
Neurology	South East GLH
Respiratory	South East GLH
Skin	South East GLH
Endocrinology	South West GLH
Eyes	North Thames GLH
Hearing	North Thames GLH
Immunology	North Thames GLH
Inherited Cancer	North Thames GLH
Metabolic	North Thames GLH
Mitochondrial	North Thames GLH
Musculoskeletal	North Thames GLH and Central & South GLH
NIPD	North Thames GLH
Renal	North Thames GLH and North East & Yorkshire GLH