Sheffield Diagnostic Genetics Service

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Dear Colleagues

Re: Update on delays to Genomic Laboratory testing services

I am writing to update you on the ongoing delays to the Genomic Laboratory testing service. These delays are for a combination of reasons centred around significant national and local reconfiguration of genomic services. This means that many of our current services are not being reported to the turnaround time guidelines that you may normally expect.

This is affecting services at each of our Genomic Laboratories in the North East and Yorkshire Genomic Laboratory Hub (NEY GLH). We continue to work together as the NEY GLH to manage recovery plans across the wider patch to rectify this and improve performance. However it will take several months before all turnaround times return to the expected levels.

Please be assured that we will prioritise the most urgent clinical work which includes:

- pregnant women undergoing prenatal diagnosis;
- patients needing urgent advice on carrier testing relating to pregnancy, for example cystic fibrosis, thalassaemia;
- those faced with abnormal fetal scans; critically ill neonates and children requiring assessment and those for whom the rapid PICU/NICU WES is appropriate;
- conditions where rapid genetic testing may alter clinical treatment or decision making; and
- patients requiring urgent testing, for example BRCA testing, to inform chemotherapy options.

On the next page, we have provided a set of indicative turn-around times to help manage service expectations as we balance the capacity and build recovery.

Sheffield

Test	Indicative laboratory reporting time
ONCOLOGY	
DPYD	3 - 7 days
BCR-ABL and chimerism monitoring	13 - 23 days
JAK2/MPN	8 - 28 days
Myelodysplastic syndrome	21 - 46 days
WGS	3 - 6 months
RARE DISEASE	
SNP microarrays (postnatal bloods)	5 – 10 weeks
Karyotyping (urgent)	5 - 7 weeks
Karyotyping (routine)	4 – 6 months
R184/R185 Cystic fibrosis testing	2 – 5 weeks
R134 Familial hypercholesterolaemia	5 - 7 weeks
R240/R242/R244 targeted familial testing	6 – 10 weeks
R95 Hereditary Haemochromatosis	5 - 7 weeks
R207/R208 Inherited breast and/or ovarian cancer	5 – 9 weeks
Urgent sickle cell/thalassaemia screening/antenatal testing	2 – 4 weeks
Non-urgent sickle cell/thalassaemia testing	10 – 12 months
Haemophilia/haemostasis single gene testing	10 – 12 months
Small/medium NGS panels	6 – 9 months
Large NGS panels	11 – 13 months
WGS panels (Core)	7 – 10 months
WGS panels (Neurology)	11 – 15 months

In order to help us manage our services, if you are uncertain whether a genetic test is suitable please do not hesitate to contact your local genomic laboratory before sending a sample.

We wish to thank you for your understanding and cooperation.

Yours sincerely

Richard Kirk

Richard Kirk MSc FRCPath
Head of Department