All Wales Medical Genomics Service
Institute of Medical Genetics
Bwrdd Iechyd Prifysgol Caerdydd a'r Fro
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All Wales Medical Genomics Service
Institute of Medical Genetics
Cardiff and Vale University Health Board
University Hospital of Wales
Heath Park Cardiff
CF14 4XW

Reception ☎ (029) 2184 4023

5st November 2024

Dear Service User,

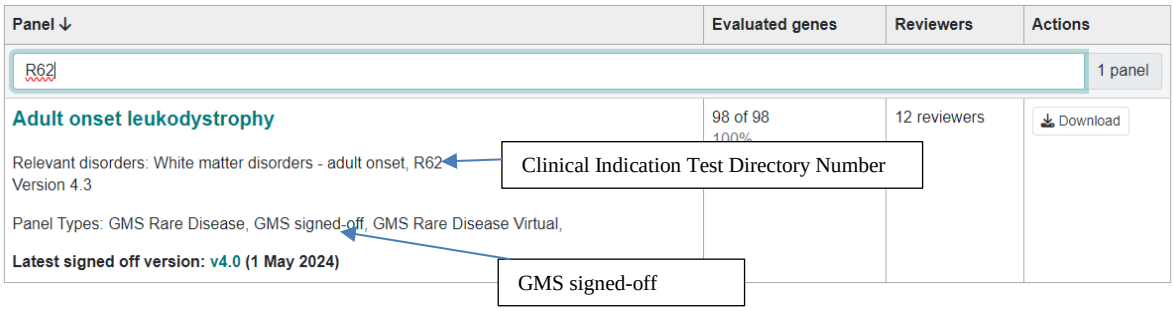
Re: Changes to providers of testing and associated documentation

It has recently become necessary to change the external laboratory supplier for a number of Rare Diseases Test Directory tests, please see reverse for details.

These tests will be equivalent to those previously provided but you will notice some changes to how you request the test and to the report format. This leaflet provides some important information to support with these test requests.

1) Test information

The test you request will be a singleton, whole exome sequencing (WES) based test of a panel of genes that are directly related to the phenotypic presentation in the patient. The genes that will be included in the panel of genes tested can be found on the PanelApp website [Panels (genomicsengland.co.uk)] and will include analysis of 'green' genes only. Please note the panel test is validated for small sequence variants only e.g. single nucleotide variants (SNVs) and multinucleotide variants (InDels). Copy number variants (CNVs), short tandem repeats (STRs) and genes encoded by the mitochondrial genome are not included in the analysis as the panel is NOT delivered by whole genome sequencing (WGS). However, CNVs may be reported if they are detected. STRs listed in the test directory panels are not included and should be managed as separate test requests if required based on the clinical phenotype in your patient. The panel version used will be a 'GMS signed-off' version as this denotes the gene list and version of the panel that has been approved for use as part of the 'genomic medicine service' as opposed to some other research-based panels that are also included in PanelApp. Test details will be included on the report.



2) Test Request form

In order to initiate tests for the relevant indications with the external supplier, please complete the associated WES request form stating the clinical details and confirming that the patient meets the clinical criteria listed in the NHSE Rare and inherited disease eligibility criteria document [REX RD (Word) (england.nhs.uk)]. Reports will be received by the AWMGS laboratory and sent on to the referring clinical team by post and by e-mail. Please ensure that the email address you would like the finalised report returned to is included on the form.

Electronic versions of this letter and WES request form are available on the AWMGS website: select the 'Health Professional' information tab, then 'Which genetics test do you require' option and search for the R# or test name in the search field; documents are available from the 'Download' column.

I hope this information is useful but if you have any further queries please contact lab.genetics.cav@wales.nhs.uk and we will respond as quickly as we are able.

Best wishes,

Sian Corrin

Rare Diseases Service Lead

All Wales Medical Genomics Service

AWMGS

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Test/panel name	Test Directory 'R' code	Test methodology
R54 Hereditary ataxia with onset in adulthood	R54.3	WES gene panel
R55 Hereditary ataxia and cerebellar anomalies - childhood onset	R55.4	WES gene panel
R56 Adult onset dystonia, chorea or related movement disorder	R56.3	WES gene panel
R57 Childhood onset dystonia, chorea or related movement disorder	R57.5	WES gene panel
R58 Adult onset neurodegenerative disorder	R58.4	WES gene panel
R60 Adult onset hereditary spastic paraplegia	R60.3	WES gene panel
R61 Childhood onset hereditary spastic paraplegia	R61.4	WES gene panel
R62 Adult onset leukodystrophy	R62.2	WES gene panel
R86 Hydrocephalus	R86.3	WES gene panel
R381 Other rare neuromuscular disorders	R381.2	WES gene panel
R15 Primary immunodeficiency or monogenic inflammatory bowel disease	R15.5	WES gene panel