

How to use PanelApp



The National Genomic Rare and Inherited Disease Test Directory indicates that gene panels are the appropriate test for many clinical indications. The gene panels signed off for use within the Genomic Medicine Service (GMS) are listed in PanelApp.

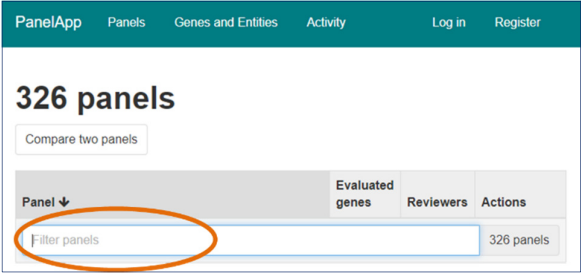
PanelApp is a crowdsourcing tool to allow gene panels to be shared, downloaded, viewed and curated by the scientific and clinical community.

To determine which genes are on the gene panel for a test associated with a clinical indication you can go to the PanelApp website and follow the steps below:

<https://panelapp.genomicsengland.co.uk/>

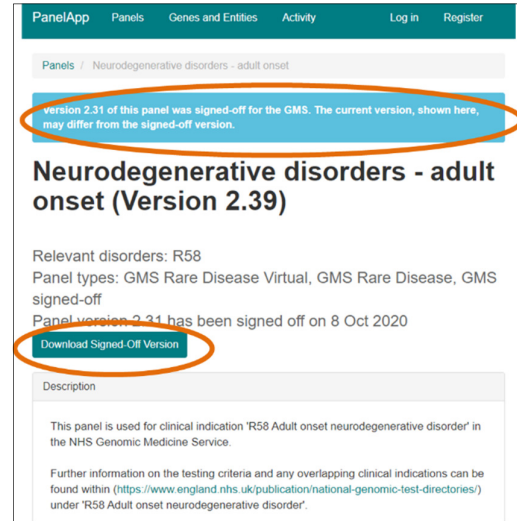
Searching for Gene Panels

- 1) Use the Rare and Inherited Disease Test Directory to identify the clinical indication
- 2) On the opening web page select Panels in the menu bar at the top of the page



- 3) Search for clinical indication or R code
or
Search using the term 'GMS' to browse the panels associated with the GMS Test Directory

- 4) The blue panel indicates that this panel is a GMS Panel associated with the Test Directory
- 5) Download the GMS signed off version of the gene panel by selecting the green box

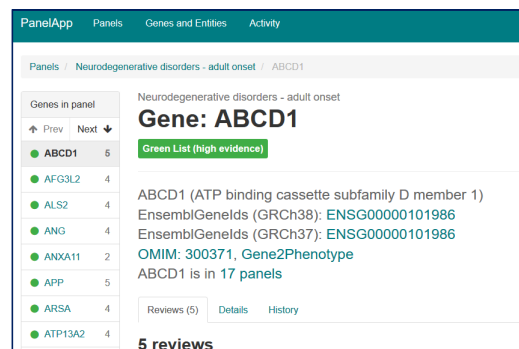


List	Entity	Reviews	Mode of inheritance	Details
	Filter Entities			422 Entities
Green	ABCD1	5 reviews 2 green	X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males)	Sources <ul style="list-style-type: none"> Expert list Expert Review Green London North GLH NHS GMS Wessex and West Midlands GLH Yorkshire and North East GLH Phenotypes <ul style="list-style-type: none"> Hereditary spastic paraplegia adrenal failure VLCFA accumulation spastic paraparesis Tags
Green	AFG3L2	4 reviews 3 green	BOTH monoallelic and biallelic, autosomal or pseudautosomal	Sources <ul style="list-style-type: none"> Expert Review Green London North GLH NHS GMS Wessex and West Midlands GLH Yorkshire and North East GLH Phenotypes <ul style="list-style-type: none"> Spinocerebellar ataxia 28 Spinocerebellar Ataxia, Dominant Ataxia, spastic, 5, autosomal recessive Dystonia Spastic ataxia 5, autosomal recessive Tags


- 6) Browse all the genes on a panel by scrolling down the page

(but note that this panel version may not be the GMS signed off panel version)

- 7) Selecting each gene provides additional details and the gene-disease curation evidence



PanelApp Gene Ratings



RED **STOP:** not enough evidence for this gene-disease; this gene should not be used for genome interpretation.

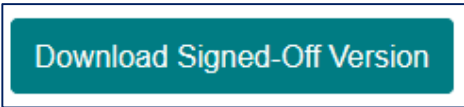
AMBER **PAUSE:** moderate evidence for this gene-disease association, and should not yet be used for genome interpretation.

GREEN **GO:** high level of evidence for this gene-disease association, demonstrates confidence that this gene should be used for genome interpretation.

Genes in panels are classified according to a traffic light system based on the evidence supporting the gene-disease association

Only Green genes will be analysed and reported for a GMS panel test

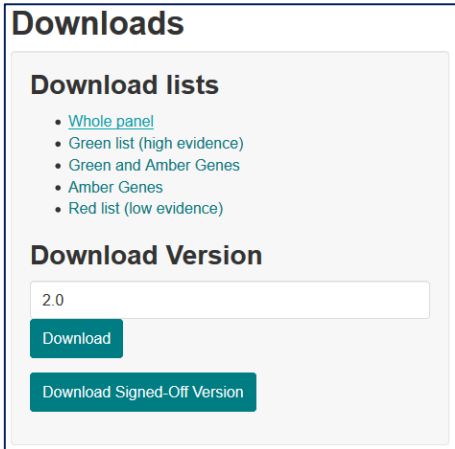
Downloading Gene Panels



Download the GMS signed off version of a gene panel by selecting the green box

To download a previous version of a panel scroll down to the bottom of the panel webpage

Add the panel version number to the search box – for example “2.0” – and select “Download”



Downloads

Download lists

- [Whole panel](#)
- [Green list \(high evidence\)](#)
- [Green and Amber Genes](#)
- [Amber Genes](#)
- [Red list \(low evidence\)](#)

Download Version

2.0

Download

Download Signed-Off Version

A downloaded gene panel file can be opened in excel

The file contains multiple columns with gene specific information

	O	P	Q	R	S	M
1	Flagged	GEL_Status	UserRatings_Green	version	ready	
2			3		3.2	
3			3		3.2	
4			3		3.2	
5			3		3.2	
6			3		3.2	
7			3		3.2	
8			3		3.2	
9			3		3.2	
10	72503;792		3		3.2	
11			3		3.2	O
12			3		3.2	

Useful columns:

Column P - GEL_Status:

- 3 = Green Gene
- 2 = Amber Gene
- 1 = Red Gene

Column R - Version

Version of the downloaded gene panel

Useful web links

National Genomic Rare and Inherited Disease Test Directory:

<https://www.england.nhs.uk/publication/national-genomic-test-directories/>

PanelApp Website:

<https://panelapp.genomicsengland.co.uk/>

Become a reviewer to curate PanelApp gene content:

<https://panelapp.genomicsengland.co.uk/#!Reviewers>