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

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

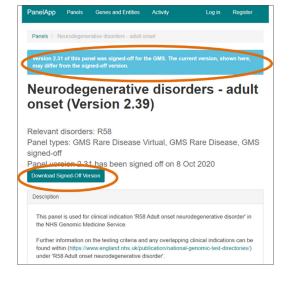


PanelApp	Panels	Genes and Entities	Activity	Log in	Register
326 p	anel	S			
Compare two	panels				
Panel 🗸			Evaluated genes	Reviewers	Actions
Filter panels	S	>			326 panels

 Search for clinical indication or R code or

> Search using the term 'GMS' to browse the panels associated with the GMS Test Directory

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

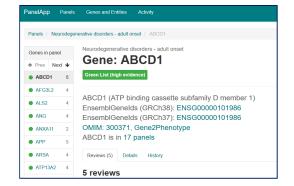


List 🛧	Entity	Reviews	Mode of inheritance	Details	
Filter En	tities				422 Entities
Green	ABCD1	5 reviews 2 green	X-LINKED: homizygous mutation in males, monoallolic mutations in females may cause disease (may be less severe, later onset than males)	Sources Egent Ist Egent	
Green	AFG3L2	4 reviews 3 green	BOTH monoallelic and biallelic, autosomal or pseudoautosomal	Source - Eigent Review Green - London North GLH - Nr6 Guills - Nr6 Source and North East GLH - Noncorrebellar allowa GLH - Spinocerebellar allowa GL - Spinocerebellar Allowa Dominant - Allawa, spasit, d. autosomar recessive - Spisocare allowa da and a subsonai recessive - Spisocare allowa da a subsonai recessive - Spisocare allowa da a subsonai recessiv	

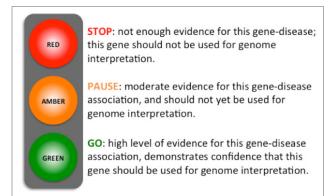
 Browse all the genes on a panel by scrolling down the page

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

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



PanelApp Gene Ratings



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



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"

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

Downloads	
Download lists	
 <u>Whole panel</u> Green list (high evidence) Green and Amber Genes Amber Genes Red list (low evidence) 	
Download Version	
Download	
Download Signed-Off Version	



A downloaded gene panel file can be opened in excel

The file contains multiple columns with gene specific information

41			∫₂ Er	tity Name																
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5 ACTB	gene	ACTB		eintellect				Dystonia				22366783	25525582,2	4896178	3		3.2			ENSGO
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7 ACTL64		ACTLEA		eintellect				L develop				28649782			3		3.2			ENSGO
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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