

A Reviewers' Guide To PanelApp



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Reviewing Panels in PanelApp

- Expert review of the gene panels is sought to enable a community consensus to be reached on which genes and genomic entities should appear on a diagnostic-grade panel for each disorder.
- We request that reviewers have expertise in a disease area relevant to the panel they are reviewing.
- Reviewers can be based anywhere in the world, and can have an academic, clinical or commercial background.
- This guide highlights the key **Review** functions of PanelApp, in a series of how-to steps. The guide can be used alongside the current PanelApp handbook, which details how to browse PanelApp and leave reviews.
- The accompanying handbook is linked from the Genomics England PanelApp page: https://panelapp.genomicsengland.co.uk/



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Creating a PanelApp Reviewer Account, and Logging in





Register

Username

Username

Required. 150 characters or fewer. Letters, digits and @/./+/-/_6n

First name

First name

Last name

Last name

Email address

Email address

Confirm email

Password

- Your password can't be too similar to your other personal information.
- Your password must contain at least 8 characters.
- Your password can't be a commonly used password.
- Your password can't be entirely numeric.

Password confirmation

Password confirmation

Enter the same password as before, for verification.

Affiliation

Affiliation

Role

Please select a role

Workplace

Please select a workspace

Group

Please select a group

Your Username and password are <u>not visible</u> to the public or other users.

• Please choose a username without spaces, and do not use your email as your username.

Please use your institutional/work/NHS email address to register.

Fill in your details on the Registration form, and click 'Register'.

• After registering, you will be sent an email requiring you to verify your email address by visiting a link provided in the email; note that the link expires in 3 days.

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- Please click on this link to enable your registration to be processed.
- Your application will be reviewed by a Genomics England curator within 48 hours. If accepted, you will be sent a confirmation email and then you can log in to PanelApp to provide reviews on panels.
- While your application is being processed, you can still browse PanelApp as a public user.
- Your Affiliation, Role and Workplace are collected to help verify your Reviewer account.
- Your Workplace and Group will be displayed on PanelApp.



<u>Note</u>: Once registered, if you forget your password, you can reset your account by selecting **I forgot my password** on the PanelApp login page (<u>https://panelapp.genomicsengland.co.uk/accounts/login/</u>) then enter the same email you registered with, and select **Request new password**.





Finding your panel or gene of interest in PanelApp

Use the top **PanelApp Toolbar** to log in to your reviewer account, and search for your panel or gene of interest:





Please note that we have changed the names of some of our panels - you will still be able to search for the previous name via <u>the 'panels' page</u> or <u>via our webservices/API</u>, as this will be added as a 'relevant disorder' to the panel. The panel code (identified in the URL) will remain the same.

https://panelapp.genomicsengland.co.uk



PanelApp Panels Genes and Entities Activity rebecca_reviewer Log out Clicking on **Panels** in the top Toolbar will list all panels. Type in 293 panels the Filter panels box to find your panel of interest. Compare two panels Panel 🕹 **Evaluated genes** Reviewers Actions 3 panels limb VACTERL-like phenotypes Click on a panel name to: viewers Lownload View the panel description. Level 3: Limb disorders 1) Level 2: Dysmorphic and congenital abnormality View the panel type. 2) 3) View Genes and Genomic Entities on the Relevant disorders: Version 1 22 panel, and their current ratings. Select a gene on the panel to review. 4) Limb girdle muscular dystrophy viewers Download Level 3: Neuromuscular disorders Each panel is versioned. Level 2: Neurology and neurodevelopmental dis Version 0 panels have not yet been finalised, and are not used in the Version 1.12 interpretation pipeline. Limb disorders Version 1 and Version 2 panels have been reviewed, validated by a curator, Version 1.2 and are used in the interpretation pipeline. Each change to a panel increases the minor version incrementally (e.g. Version 1.11 to Version 1.12. Note that Version 1.12 of a gene panel is more 18/12/18 recent than Version 1.2.



An overview of information captured on a PanelApp panel

Panel View in PanelApp:



PanelApp Genes and Entities

Panels / Limb disorders

Limb disorders (Version 1.3)

Panel types: Rare Disease 100K, GMS Rare Disease Virtual

This panel is a virtual panel that can form part of the analysis of a broader phenotype, where relevant, using genome or exome data in the NHS Genomic Medicine Service. This is not a primary panel for any GMS clinical indications. Ellen McDonagh (Genomics England) The content of this panel is overseen by NHS Genomic Medicine Service governance. Group: other This panel was originally developed for the 100,000 Genomes Project and is still being used for participants in the project. For the rare disease eligibility criteria refer to: https://www.genomicsengland.co.uk/rarediseasecriteria100K Richard Scott (Genomics England) This gene panel is designed to cover the following limb disorders, where limb disorder is the primary or secondary feature : Ana Beleza (Guy's and St Thom Foundation Trust) Group: Other NHS organisation Workplace: NHS clinical service Olivia Niblock (Genomics England) Olivia Niblock (Genomics England) Prodydactyly (HP:0001156) Group: Other NHS organisation Strondactyly (HP:0001156) Group: Other NHS organisation Pholydactyly (HP:0001156) Group: Other NHS organisation Pholydactyly (HP:0001156) Group: Other Vorkplace: NHS clinical service Olivia Niblock (Genomics England) Olivia Niblock (Genomics England) Group: Other Workplace: Other Sarah Leigh (Genomics England)	Description	12 reviewers
This panel was originally developed for the 100,000 Genomes Project and is still being used for participants in the project. For the rare disease eligibility criteria refer to: https://www.genomicsengland.co.uk/rarediseasecriteria100K Richard Scott (Genomics England This gene panel is designed to cover the following limb disorders, where limb disorder is the primary or secondary feature : Ana Beleza (Guy's and St Thom Foundation Trust) Brachydactyly (HP:001156) Group: Other WHS organisation - Dilgodactyly (HP:001156) Group: Other WHS organisation - Syndactyly (HP:0012165) Group: Other WHS organisation - Polydactyly (HP:001042) Olivia Niblock (Genomics England - Annelia (HP:0009827) Olivia abnormalities - Ullnar ray abnormalities - Badial ray abnormalities (HP:0410	This panel is a virtual panel that can form part of the analysis of a broader phenotype, where relevant, using genome or exome data in the NHS Genomic Medicine Service. This is not a primary panel for any GMS clinical indications. The content of this panel is overseen by NHS Genomic Medicine Service governance.	Ellen McDonagh (Genomics Eng Curator) Group: other Workplace: other
This gene panel is designed to cover the following limb disorders, where limb disorder is the primary or secondary feature : Ana Beleza (Guy's and St Thom Foundation Trust) - Brachydactyly (HP:0001156) Group: Other NHS organisation - Dilgodactyly (HP:0100257) Group: Other NHS organisation - Polydactyly (HP:0001159) Group: Other NHS organisation - Polydactyly (HP:0001159) Group: Other NHS organisation - Polydactyly (HP:0001159) Group: Other NHS organisation - Polydactyly (HP:000142) Group: Other - Annelia (HP:0009827) Group: Other - Ulnar ray abnormalities Group: Other - Radial ray abnormalities (HP:0410 Sarah Leigh (Genomics England)	This panel was originally developed for the 100,000 Genomes Project and is still being used for participants in the project. For the rare disease eligibility criteria refer to: https://www.genomicsengland.co.uk/rarediseasecriteria100K	Group: Other Workplace: Genomics England
- Syndactyly (HP:0001159) - Polydactyly (HP:0010442) - Amelia (HP:0009827) - Phocomelia (HP:0009829) - Ulnar ray abnormalities - Radial ray abnormalities (HP:0410 Sarah Leigh (Genomics England	This gene panel is designed to cover the following limb disorders, where limb disorder is the primary or secondary feature : - Brachydactyly (HP:0001156) - Oligodactyly (HP:000257) - Ectrodactyly (HP:00257)	Ana Beleza (Guy's and St Thoma Foundation Trust) Group: Other NHS organisation Workplace: NHS clinical service
- Ulnar ray abnormalities - Radial ray abnormalities (HP:0410 Sarah Leigh (Genomics England	- Syndactyly (HP:0001159) - Polydactyly (HP:001042) - Arnelia (HP:0009827) - Phocomelia (HP:0009829)	Olivia Niblock (Genomics Englar Group: Other Workplace: Other
	- Ulnar ray abnormalities - Radial ray abnormalities (HP:0410	Sarah Leigh (Genomics England

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Log in Register

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Panel types denote the project(s) the panel is used for, and include:

- Rare Disease 100K: a panel used for the interpretation pipeline for rare disease genomes from the 100,000 **Genomes Project**
- Cancer Germline 100K: a panel used for the • interpretation pipeline for cancer germline genomes from the 100,000 Genomes Project
 - **GMS Rare Disease**: a panel developed for the NHS Genomic Medicine Service - may be delivered by WES, large or small 'wet lab' panel or as a virtual panel GMS Rare Disease Virtual: a panel developed for the
 - NHS Genomic Medicine Service will be used as a virtual panel for whole genome sequencing indications

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Green ARSE 2 reviews X-LINKED: hemizygous Sources	
Add review 1 green Add review and the set of the set o	
Oreen BHLHA9 3 reviews BIALLELIC, autosomal or pseudoautosomal • Dispert Review Green Ovide 1 green I green • Usert Review Green • Viapath • Viapath • Viapath • Vidorian Clinical Genetics Services • Syndactyly, mesoaxial synostotic, with phalangeal reduction, 09432, • Polydactyly	
ing s	

Please read the panel Description box before leavin review. The Description box contains key informatio which can include:

- **Clinical indication**
- Governance

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- Details on how the panel was created
- **Technical considerations** •

Click on a gene symbol to se further gene details, and pr a review.





Understanding Gene Ratings in PanelApp





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

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

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

We classify genes on a panel according to a traffic light system. Genes are rated in terms of the level of evidence to support their association with the phenotypes covered by the gene panel in question.

- Reviewers are asked to rate genes according to this traffic light system.
- Green genes on Version 1+ panels will reflect this evidence system and can be used for genome interpretation.

For rare disease, the criteria for assessing the evidence were developed from a combination of the ClinGen DEFINITIVE and DDG2P CONFIRMED gene evidence levels (set out in full on the next slide). In summary:

- A diagnostic-grade (Green) rating on a Version 1+ panel requires evidence from 3 or more unrelated families OR from 2-3 unrelated families where there is strong additional functional data.
- Genes that do not meet these criteria are rated as Amber (borderline) or Red (low level of evidence), and are <u>not</u> used for genome interpretation/diagnostic testing.



A. There are plausible disease-causing variants¹ within, affecting or encompassing an interpretable functional region of this gene² identified in multiple (3 or more) unrelated cases/families with the phenotype³.

OR

B. There are plausible disease-causing variants¹ within, affecting or encompassing cis-regulatory elements convincingly affecting the expression of a single gene identified in multiple (3 or more) unrelated cases/families with the phenotype³.

OR

C. As definitions A or B but in 2 or 3 unrelated cases/families with the phenotype, with the addition of convincing bioinformatic or functional evidence of causation e.g. known inborn error of metabolism with mutation in orthologous gene which is known to have the relevant deficient enzymatic activity in other species; existence of an animal model which recapitulates the human phenotype.

AND

D. Evidence indicates that disease-causing variants follow a Mendelian pattern of causation appropriate for reporting in a diagnostic setting⁴.

AND

E. No convincing evidence exists or has emerged that contradicts the role of the gene in the specified phenotype.

¹*Plausible disease-causing variants: Recurrent de novo variants convincingly affecting gene function. Rare, fully-penetrant variants - relevant genotype never, or very rarely, seen in controls.*

²Interpretable functional region: ORF in protein coding genes miRNA stem or loop.

³*Phenotype: the rare disease category, as described in the eligibility statement.*

⁴Intermediate penetrance genes should not be included.

Adapted from references: PMID:28552198 and PMID: 25529582



Leaving a Review in PanelApp

When reviewing a gene or genomic entity, please **rate** whether there is sufficient evidence for the gene or genomic entity to be on a diagnostic panel.

You can also **add the following fields** when reviewing, although they are not compulsory, they are useful when a curator is collating the reviews.

- Mode of inheritance
- Mode of pathogenicity
- Publications
- Phenotypes
- Free-text comments





Genes in panel				
Prev	Next	≁		
ARHG	AP31	3		
ARSE		2		
BHLH/	49	3		
BMPR	1B	2		
BRCA	2	0		
BRIP1		0		
DLX5		3		
OOCK	6	2		
OVL1		3		
EBP		1		
EOGT		2		
ERCC	4	0		
ESCO	2	0		
FAM5	8 A	4		
FANC	A	0		
FANC	в	0		
FANC	С	0		
FANC	D2	0		
FANC	E	0		
FANC	F	0		





If the mode of inheritance you want is not within the dropdown menu, select 'Other' and provide details in the comments box.

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If known, please provide information regarding imprinting by selecting either the maternally imprinted or paternally imprinted mode of inheritance, and leaving details in the comments box.

Definitions for each mode of inheritance term can be found by clicking on the question-mark pop-up icon.

MONOALLELIC, autosomal or pseudoautosomal, maternally imprinted (paternal allele expressed)

MONOALLELIC, autosomal or pseudoautosomal, paternally imprinted (maternal allele expressed)

MONOALLELIC, autosomal or pseudoautosomal, imprinted status unknown

BIALLELIC, autosomal or pseudoautosomal

BOTH monoallelic and biallelic, autosomal or pseudoautosomal

BOTH monoallelic and biallelic (but BIALLELIC mutations cause a more SEVERE disease form), autosomal or pseudoautosomal

X-LINKED: hemizygous mutation in males, biallelic mutations in females

X-LINKED: hemizygous mutation in males, monoallelic mutations in females may cause disease (may be less severe, later onset than males) **MITOCHONDRIAL**

Unknown

Other - please specifiy in evaluation comments

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Genes in pa	anel		Limb disorders	
↑ Prev 1	Next	¥		Genomics
ARHGAR	P31	3	Green List (high evidence)	england
ARSE		2	2	
BHLHA9)	3	ARSE (arylsulfatase E (chondrodysplasia punctata 1)) ³ Ensemble analds (CRCh38): ENSC00000157399	• - •
BMPR1E	в	2	 ² EnsemblGenelds (GRCh37): ENSG00000157399 	
BRCA2		0	0 OMIM: 300180, Gene2Phenotype	
BRIP1		0	ARSE is in 6 panels	
DLX5		3	 It Ioss-ot-function variants of the phenotype please select and the phenotype ple	<u>10 not</u> cause the disease
• DOCK6		2	² P eview gene	
OVL1		3	³ Pathogenicity dropdown mo	enu.
EBP		1	Rating:	
EOGT		2	Provide rating • If providing exceptions to lo	oss-of-function, please leave a
ERCC4		0	Mode of Inheritance: free-text comment to expla	in your selection (e.g. detailing
 ESCO2 		0	Provide a mode of inheritance literature or clinical evidence	ce).
		4	Mode of pathogenicity: 0	
FAWJOA	•	4	4 Provide exceptions to loss-of-function	
FANCA		0	Mode of pathogenicity:	
• FANCE		0		
FANCC	2	0	Provide exceptions to loss-of-function	~
FANCE2	2	0		
FANCE		0	Provide exceptions to loss-of-function	
FANCE		0	⁰ Loss-of-function variants (as defined in pop up message) DO NOT cause this phenotype - please provide	e details in the comments
			Other - please provide details in the comments	

In PanelApp, we classify loss-of-function (high impact) variants as those with the sequence ontology (SO) terms:

- transcript_ablation
- splice_acceptor_variant
- splice_donor_variant
- stop_gained
- frameshfit_variant
- stop_lost
- Initiator_codon_variant









ARSE (arylsulfatase E (chondrodysplasia punctata 1)) EnsemblGenelds (GRCh38): ENSG00000157399 EnsemblGenelds (GRCh37): ENSG00000157399 OMIM: 300180, Gene2Phenotype ARSE is in 6 panels

History

Reviews (2) Details

Review gene

Rating: Provide rating Mode of Inheritance: Provide a mode of inheritance Mode of pathogenicity: Provide exceptions to loss-of-function Publications (PMID: 1234;4321):

Publications (PMID: 1234;4321)

Phenotypes (separate using a semi-colon - ;):

Phenotypes (separate using a semi-colon - ;)

Current diagnostic: ?

Add any relevant publications.

- Please provide PubMed IDs separated by a semicolon: E.g. PMID:123456;9876545
- Include publications that provide supporting evidence for your given rating, or publications refuting the gene-disorder association.
- Where the paper doesn't have a PubMed identifier, add in the publication as free text, and these will be subsequently updated by a curator.

Add in phenotypes.

- Separate phenotypes with a semi colon.
- Include relevant identifiers where possible (e.g. OMIM disease IDs and HPO terms). You can also use free text.
- E.g. Alport syndrome, 301050; Hearing Loss





- To submit a gene list, if you have a large set of genes to review or would prefer to complete your reviews off-line, please contact <u>panelapp@genomicsengland.co.uk</u> and we can provide you with an Excel file to input your reviews.
- We also welcome reviews for Short Tandem Repeats (STRs) and Copy Number Variants (CNVs). Please refer to the PanelApp handbook for details on reviewing these genomic entities. The accompanying handbook is linked from the Genomics England PanelApp page: <u>https://panelapp.genomicsengland.co.uk/</u>





Adding Genes to a PanelApp Panel

Adding a **gene** to a Panel:



Red Ready	2 reviews Add review 1 red	BIALLELIC, autosomal or pseudoautosomal	 Sources Expert Review Red Phenotypes Autosomal recessive spastic paraplegia 15 (#270700) complex form of the disease including ataxia. Pyle et al. (2015), Brain, 138, pp.276-283. Implicated in undiagnosed ataxia. 		
Red Ready	2 reviews Add review	BIALLELIC, autosomal or pseudoautosomal	Sources Radboud University Medical Center, Nijmegen UKGTN Expert Review Red Phenotypes Spinocerebellar ataxia, autosomal recessive 5 		
+ Add a Gene to this panel + Add a STR to this panel + Add a Region to this panel If any genes are missing from a panel, you can add them using the tool bar below the Entities list:					
+ Add a Gene to this pa	anel +	Add a STR to	this panel + Add a Region to this panel		

You can also use this toolbar to add a STR or a region (including CNVs) to the panel. Please refer to the PanelApp handbook for further details, or contact us at <u>panelapp@genomicsengland.co.uk</u> so we can assist you further.



Add gene to panel

Gene sym	bol:			
Source:				
Mode of p	athogenicity:	0		
Provide e	xceptions to lo	ss-of-functio	n	
Mode of ir	heritance: 👔			
Provide a	mode of inher	itance		
Penetranc	e:			
Publicatio	ns (PMID: 123	4;4321):		
Dublicatio		.42241		

Phenotypes (separate using a semi-colon - ;):

Phenotypes (separate using a semi-colon - ;)

Tags:

Rating: 🔞

Provide rating

Current diagnostic: 🔞

Current diagnostic

Comments:



Click **Add gene** when finished. Your gene will be added to the panel as **Grey**. A curator will then curate the evidence and adjust the rating to Green, Amber or Red. Start typing an HGNC Gene symbol into the top box to select your gene to add to a panel.

You must include a source of information for the gene:disease association. E.g. literature/Expert list.

You must provide a **Mode of inheritance** for the gene-disease association.

You can also add a **Mode of pathogenicity**, **Publications** and **Phenotypes.**

If penetrance is not complete, please denote using the drop down menu in the **Penetrance** field, and provide a comment.

Select a gene rating here.

The **Comments** box can be used to leave free-text information about the gene:disorder association and why the gene was added to the panel.

Any PID information should <u>not</u> be added as the comments will be publically visible.



 If you have a large set of genes and/or genomic entities to upload to PanelApp, please contact <u>panelapp@genomicsengland.co.uk</u> and a curator can assist you.





View or Edit your Evaluations

How to view or edit your Reviews:







View Changes to Panels and new reviews



PanelAp	o Panels Genes and	Entities Activity	rebecca_reviewer Log out
Activi	ty ⊤	From the	PanelApp homepage, click on the 'Activity' page
Date	Panel	Item	Activity
Filter activiti	es		3000 actions
17 Dec 2018	Intellectual disability v2.584	ATP8B1	Konstantinos Varvagiannis Deleted their comment
17 Dec 2018	Intellectual disability v2.584	ATP8B1	Konstantinos Varvagiannis commented on gene: ATP8B1: I could not find any evidence that ATP8B1 deficiency is associated with DD/ID. Kinsley et al. (2014 - PMID: 20301474) review the spectrum of the disorder. DD/ID is not among the features and not mentioned among extrahepatic manifestations. The only possibly relevant complication is vitamin E deficiency which can lead to neurologic manifestations (but not of this type).

This will display activity for all panels in PanelApp and can be filtered for date, panel name, version, gene or genomic entity name, activity type, name of Reviewer or Curator

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How to view updates on individual Panel or Gene pages:



PanelApp Panels Genes and Entities Activity	Log in	PanelApp Panels	Genes and Entities Activity Log in Register				
Panels / Limb disorders		Panels / Limb disor	ders / ARSE				
Limb disorders (Version 1.3)		Genes in panel ↑ Prev Next ↓ ● ARHGAP31 3	Limb disorders Gene: ARSE Green List (high evidence)				
Description	12 reviewers	ARSE 2 ARSE (arylsulfatase E (chondrodysplasia punctata 1))					
This panel is a virtual panel that can form part of the analysis of a broader phenotype, where relevant, using genome or exome data in the NHS Genomic Medicine Service.	Ellen McDonagh (Genomics Curator)	BHERAS 3 BMPR1B 2	 ³ EnsemblGenelds (GRCh38): ENSG00000157399 ² EnsemblGenelds (GRCh37): ENSG00000157399 				
This is not a primary panel for any GMS clinical indications.	Group: other Workplace: other	BRCA2 2	OMIM: 300180, Gene2Phenotype ARSE is in 7 panels				
This panel was originally developed for the 100,000 Genomes Project and is still being	Richard Scott (Genomics En	BRIP1 2 DLX5 3	Reviews (2) Details History				
used for participants in the project. For the rare disease eligibility criteria refer to: https://www.genomicsengland.co.uk/rarediseasecriteria100K	Workplace: Genomics England	• DOCK6 2	History Filter Activity				
This gene panel is designed to cover the following limb disorders, where limb disorder is the primary or secondary feature : - Brachydactyly (HP:0001156)	Ana Beleza (Guy's and St Th Foundation Trust) Group: Other NHS organisatio	DVL1 3 EBP 1	11 Dec 2018, Gel status: 4 Panel promoted to version 10 click on the 'History' tab and				
 Oligodactyly (HP:0012165) Ectrodactyly (HP:0100257) Syndactyly (HP:0001159) Polydactyly (HP:0010442) Amelia (HP:0009827) Bhocompile (HP:0009820) 	Workplace: NHS clinical servic	• EOGT 2	Eleanor Williams (Genomics England Cura then 'Filter Activity'				
	Group: Other Workplace: Other	ERCC4 4 ESCO2 3					
- Ulnar ray abnormalities - Radial ray abnormalities (HP:0410049)	Sarah Leigh (Genomics Engl	• FAM58A 4	5 Apr 2018, Gel status: 4 Added New Source, Added New Source, Added New Source, Set mode of inheritance Ellen McDonach (Genomics England Curator)				
Rare multisystem ciliopathy genes are not included on this panel. If a patient is suspected of having a ciliopathy, or the possibility of a ciliopathy cannot be excluded, the IDear cultivartee cilicanthe circumstance of the supercision of the circumstance of the supercision of	Group: Other Workplace: Other	FANCE 2FANCE 5	Expert Review Green was added to ARSE. Panel: Limb disorders Radboud University Medical Center, Nijmegen was added to ARSE. Panel: Limb disorders UKGTN was added to ARSE. Panel: Limb disorders Model of inheritance for gene ARSE was set to X-I UKFD: hemicroupus mutation in males biallie mutations in females.				
the 'Hare multisystem cillopathy disorders' panel (https://panelapp.genomicsengland.co.uk/panels/150/) should be applied in addition for genome analysis. Cilliopathy genes were therefore made grey (removed) on this panel.	Rebecca Foulger (Genomics curator) Group: Other Workplace: Other	• FANCC 2	to X-LINKED, nemizygous mutation in males, bialielic mutations in ternales				
Panel Activity	Louise Daugherty (Genomics E	England	This will display the relevant activity can be				
From a Panel page, click on 'Panel Acti	vity'		filtered for date, version, activity type, name of Reviewer or Curator				
18/12/18			32				

Additional Notes for Reviewers



- Your evaluations and comments will be tagged with your name and affiliation, and are public. Your name and affiliation will appear in the list of reviewers at the top of the panel.
- The date you made your reviewer will appear, along with the version of the panel you reviewed.
- You can make multiple comments for each gene or genomic entity, and edit or delete them individually.
- Changes to the rating, mode of inheritance, mode of pathogenicity and current diagnostic practice will overwrite your initial evaluation.
- Publications and phenotypes will be saved in the evaluation tool and can be added to.
- When you have reviewed a gene or genomic entity, you can see your review under the review tab along with any reviews from other experts.
- For your reviewed genes, a tick will appear in front of the gene in the Genes in panel list. A tick together with **You reviewed** text will also be added to the Reviewed column on the main panel page.
- If you have any issues, please contact us at panelapp@genomicsengland.co.uk.

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Further thanks to:

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