A quick guide to… Genomic Entities on PanelApp

8th June 2018

PanelApp Release 2.2.0

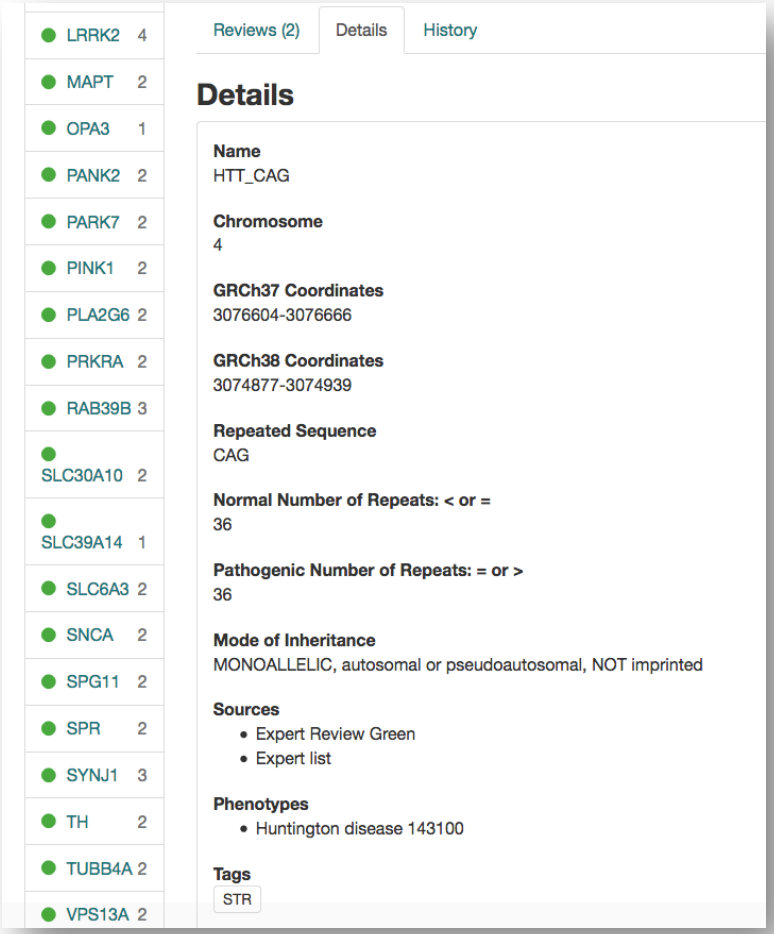
An introduction to Short Tandem Repeats (STRs) in PanelApp

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

- An interpretation pipeline for STRs in whole genome sequences has been developed and validated by Genomics England in order to allow these variant types to be reported back to Genomic Medicine Centers.
- In order to incorporate the STRs into the bioinformatics pipeline and ensure that particular STRs are only analysed in genomes for patients recruited for certain diseases, STRs have been added to the appropriate panels within PanelApp.
- In **PanelApp Release v2.2.0**, we have expanded the scope of PanelApp to allow curation and review of STRs that are disease causing when a particular number of repeats is present.
- Genes and STRs are now termed genomic 'entities' and can be added to panels, reviewed and curated.
- Green genes and Green STRs on a version 1+ panel will be used for genome interpretation for the 100,000 Genomes Project. These are deemed to have a high level of evidence for disease causation.
- **An 'Entities' page has replaced the 'Genes' page** to allow users to search for genes and STRs that are included in panels.
- **Nomenclature:** There is currently no defined nomenclature for STRs. Currently all the STRs in PanelApp are associated with a gene. We have named the STRs added to PanelApp using the associated gene symbol (HGNC-approved symbol) and nucleotide repeat - for example **HTT_CAG**.
- **How to find STRs:** STRs can be found by searching for the associated gene on the 'Entities' page. By clicking on the STR, this will take the user to the gene page where panels that include the gene or the associated STRs are displayed. From there, users can click on the panel or STR of interest.

Information provided for STRs

- Chromosome and genomic coordinates for the start and the end of the nucleotide repeats in the human reference genome for Build 37 and 38
- The repeated nucleotide sequence
- The maximum number of repeats that are considered non-pathogenic/normal (equal to or less than)
- The minimum number of repeats that are considered pathogenic/disease causing (equal to or more than)
- The mode of inheritance for the pathogenic number of repeats that is required to cause disease
- The disease caused by the pathogenic number of repeats (phenotype)
- Information about the associated gene, including links to Ensembl
- The source – “Expert list” has been used to indicate that these STRs are from the Genomics England pipeline.
- Whether penetrance is incomplete (default is complete)
- The tag ‘STR’



Example of the HTT_CAG STR in the Parkinson Disease and Complex Parkinsonism panel (version 1.40) https://panelapp.genomicsengland.co.uk/panels/39/str/HTT_CAG/#!details

How to find STRs…

PanelApp Panels Entities Activity Log in Register

Genomics England PanelApp

A crowdsourcing tool to allow gene panels to be shared, downloaded, viewed and evaluated by the Scientific Community

- Home
- News
- Navigate & Explore
- Reviewers
- Guidelines
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- FAQs
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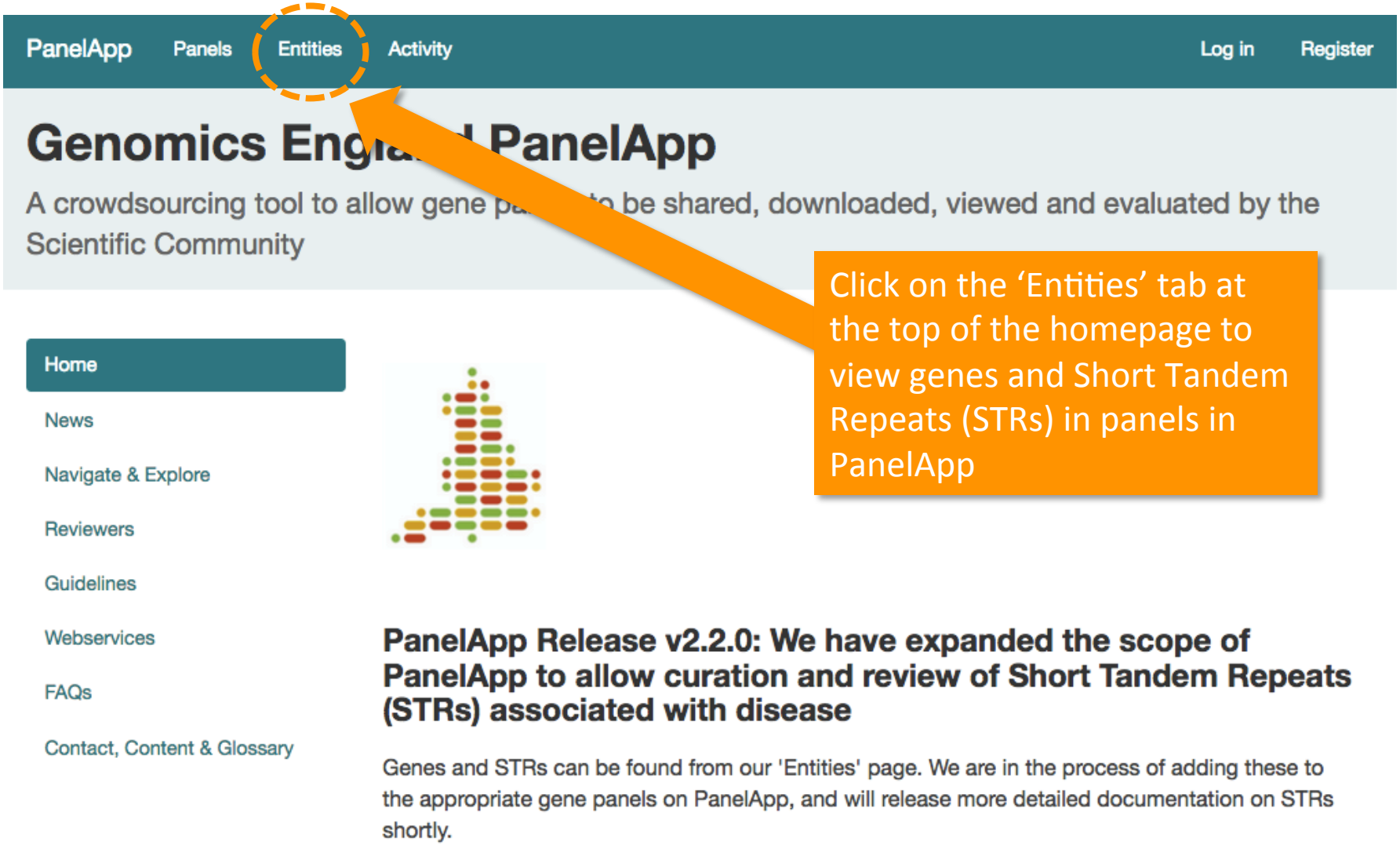
Go to

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

PanelApp Release v2.2.0: We have expanded the scope of PanelApp to allow curation and review of Short Tandem Repeats (STRs) associated with disease

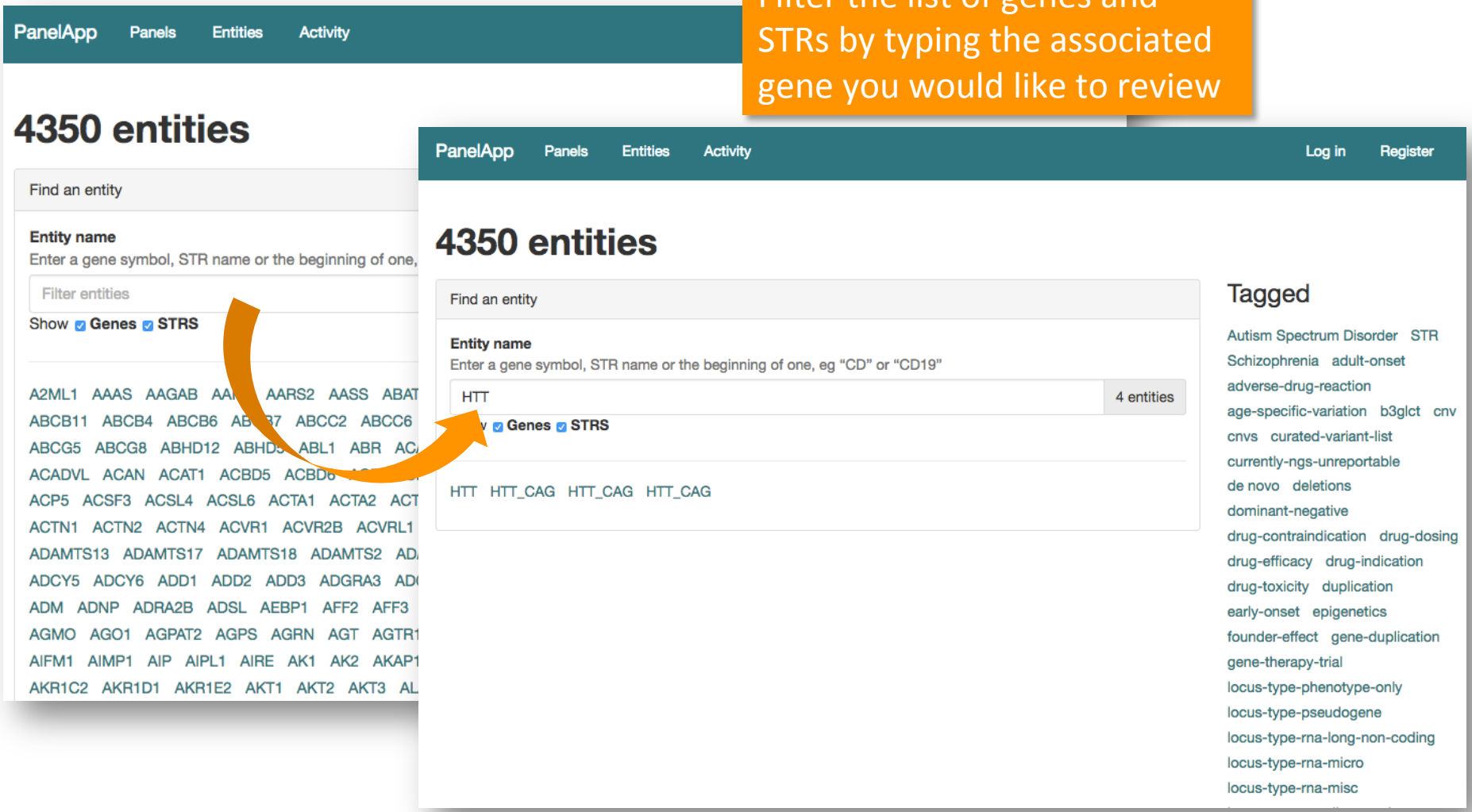
Genes and STRs can be found from our 'Entities' page. We are in the process of adding these to the appropriate gene panels on PanelApp, and will release more detailed documentation on STRs shortly.

Click on ‘Entities’

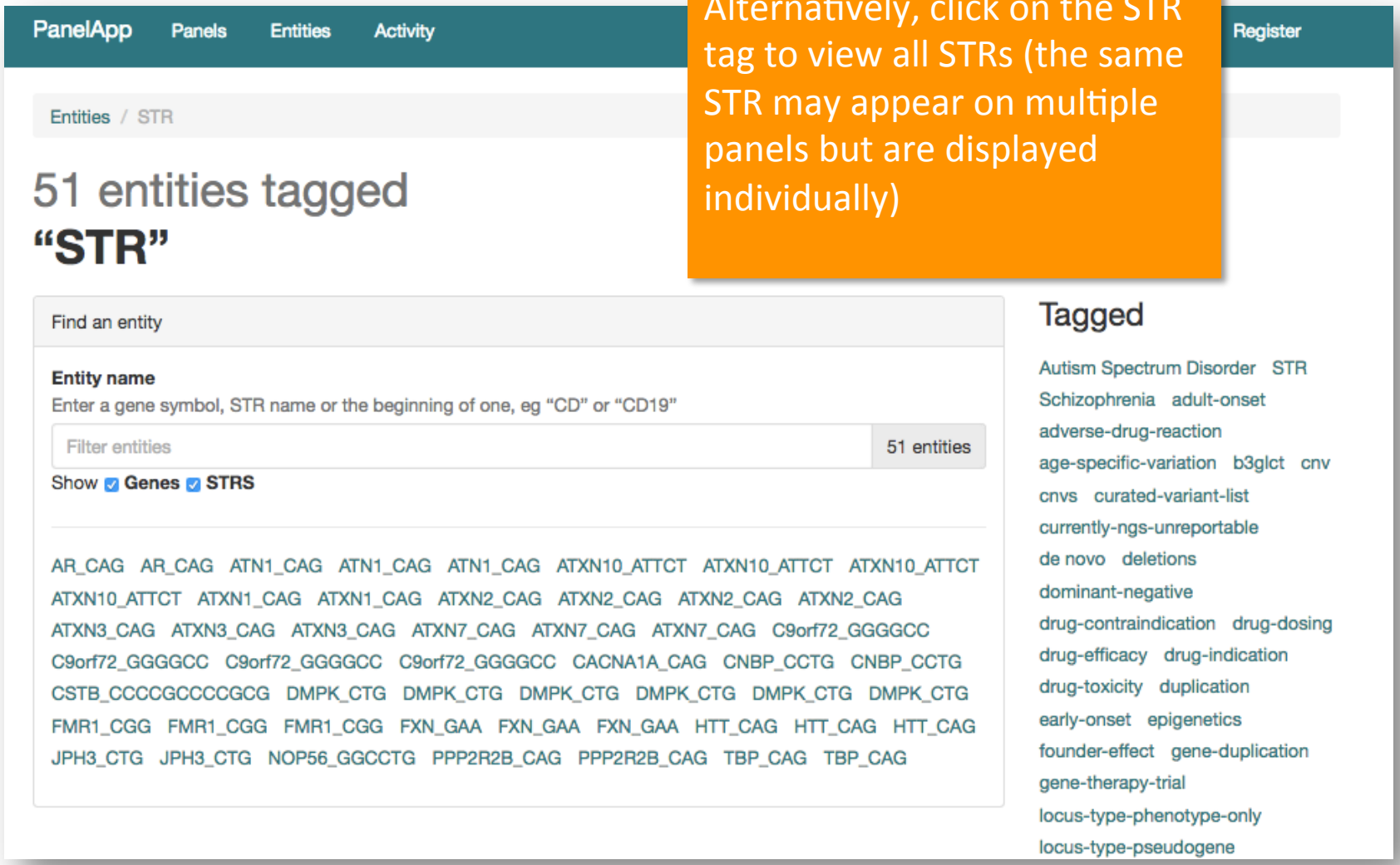


Genomic Entities page

Filter the list of genes and STRs by typing the associated gene you would like to review



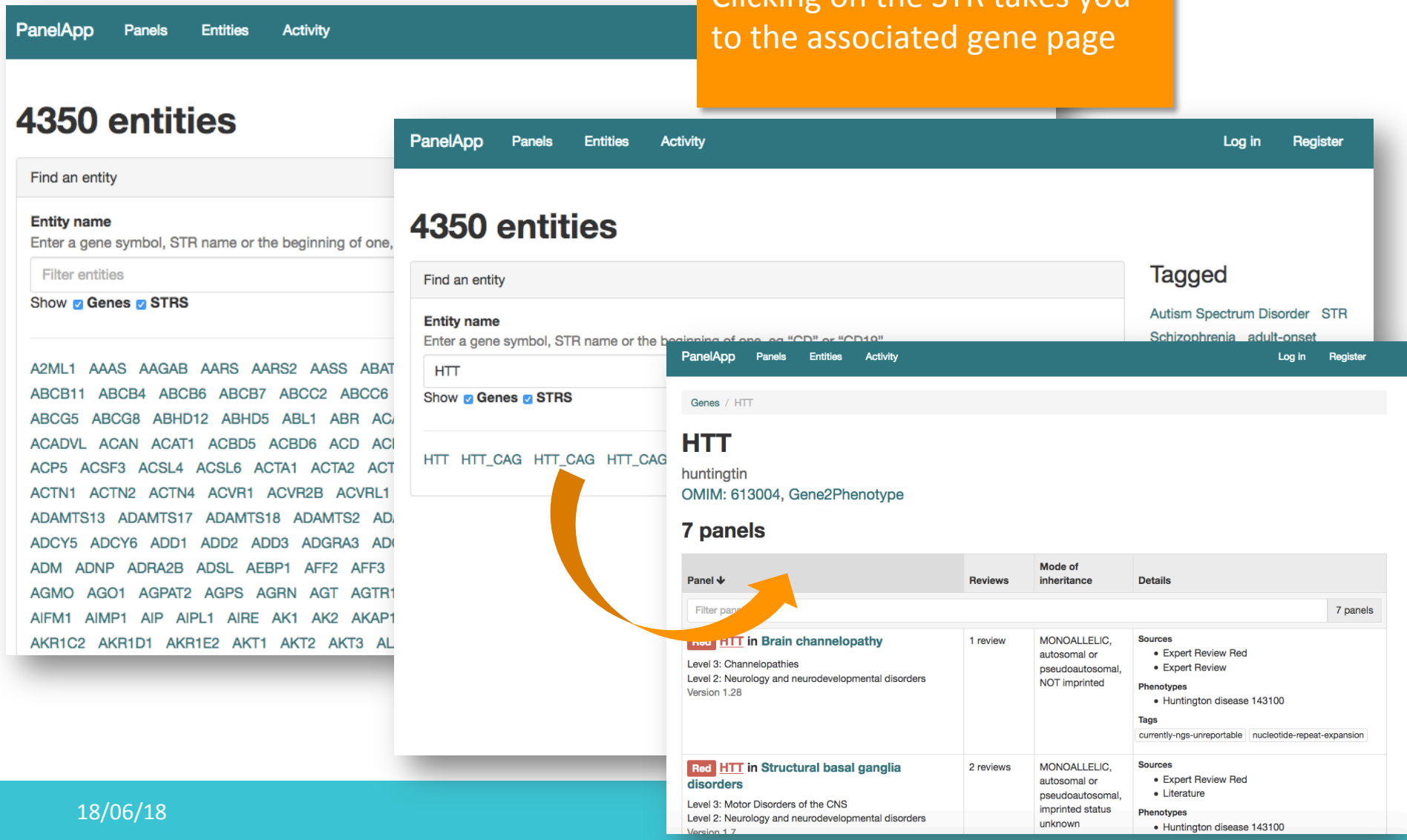
Genomic Entities page



Alternatively, click on the STR tag to view all STRs (the same STR may appear on multiple panels but are displayed individually)

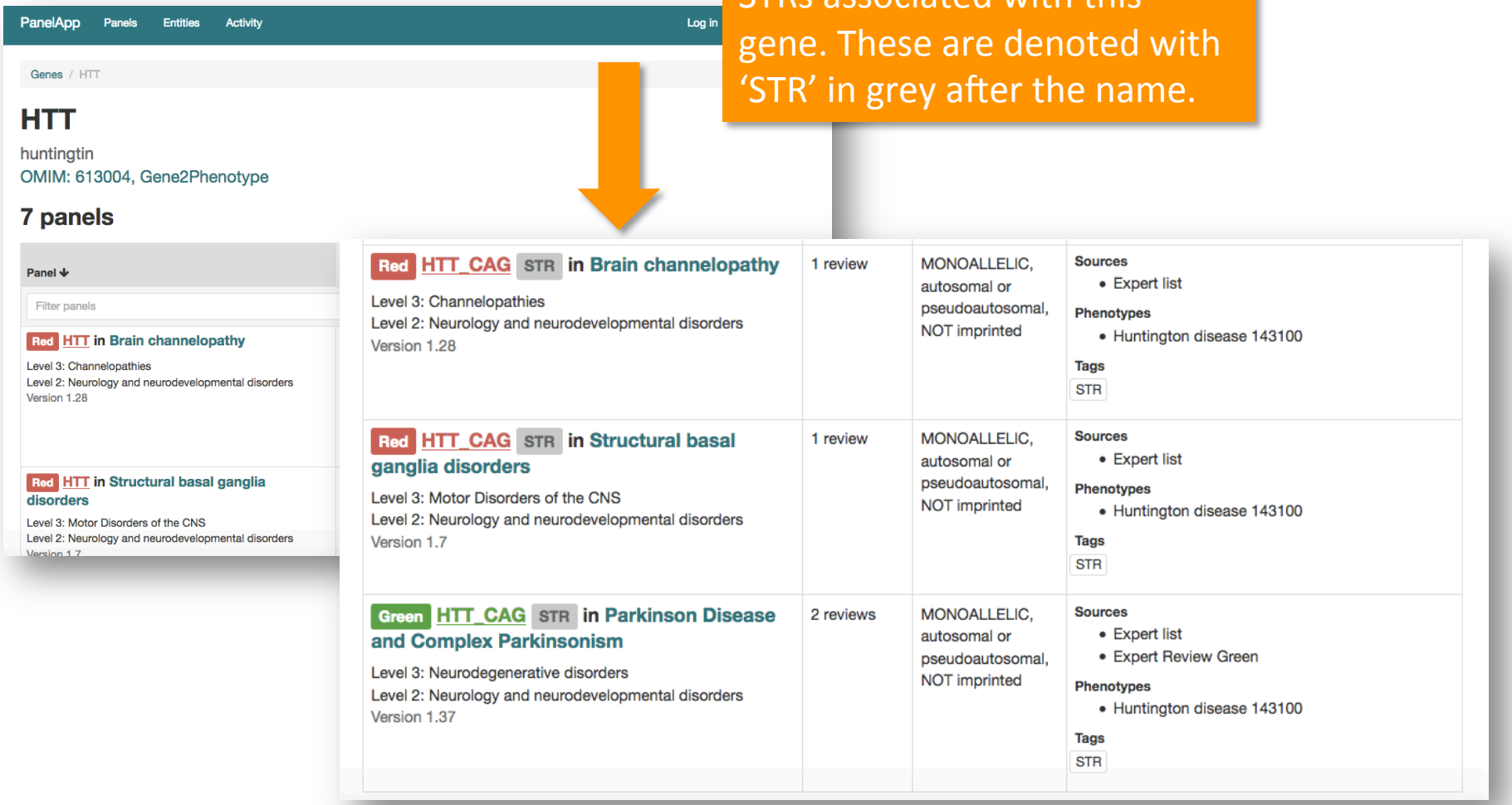
Genomic Entities page

Clicking on the STR takes you to the associated gene page



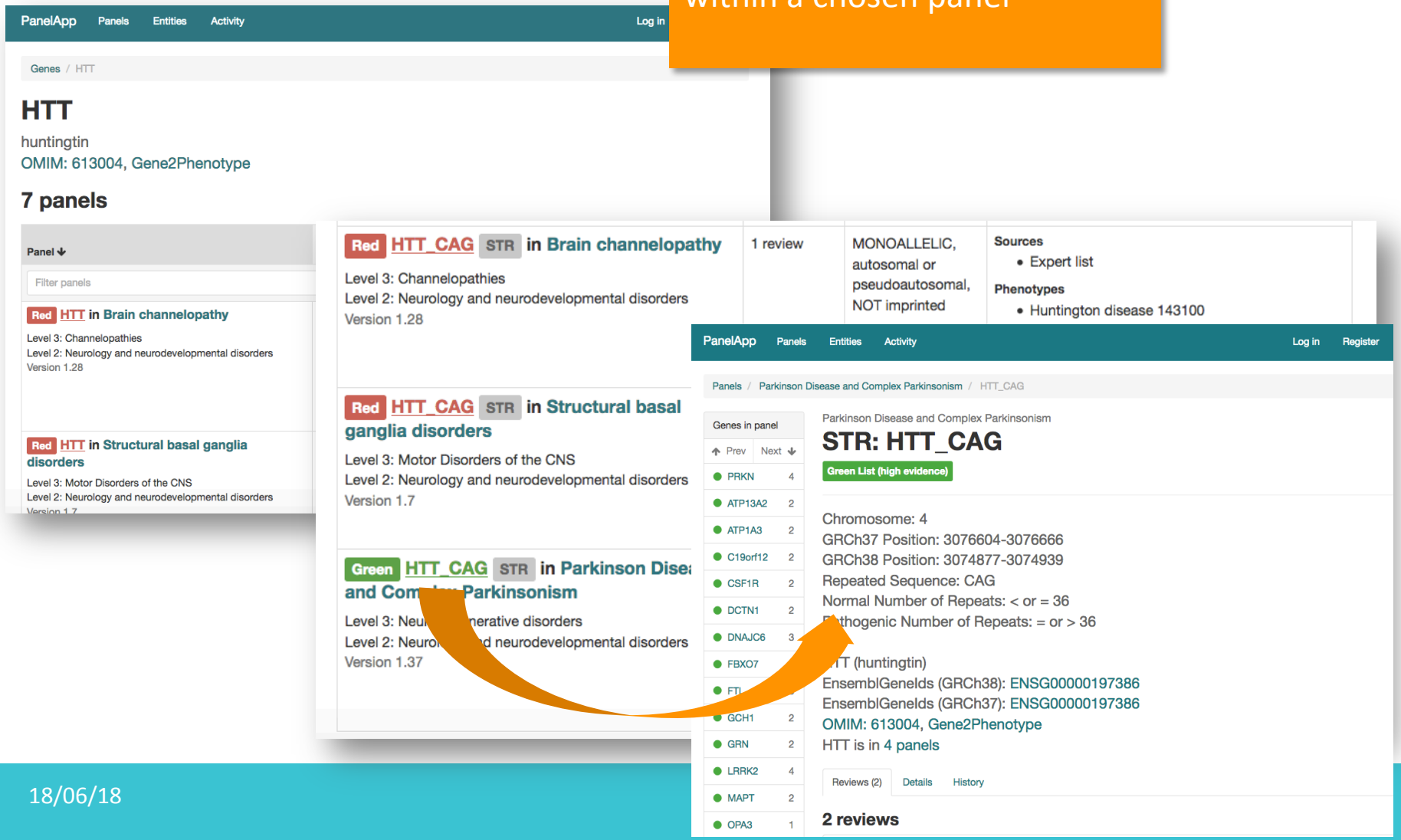
Find STRs on the associated gene page

Scroll down the page to view STRs associated with this gene. These are denoted with 'STR' in grey after the name.



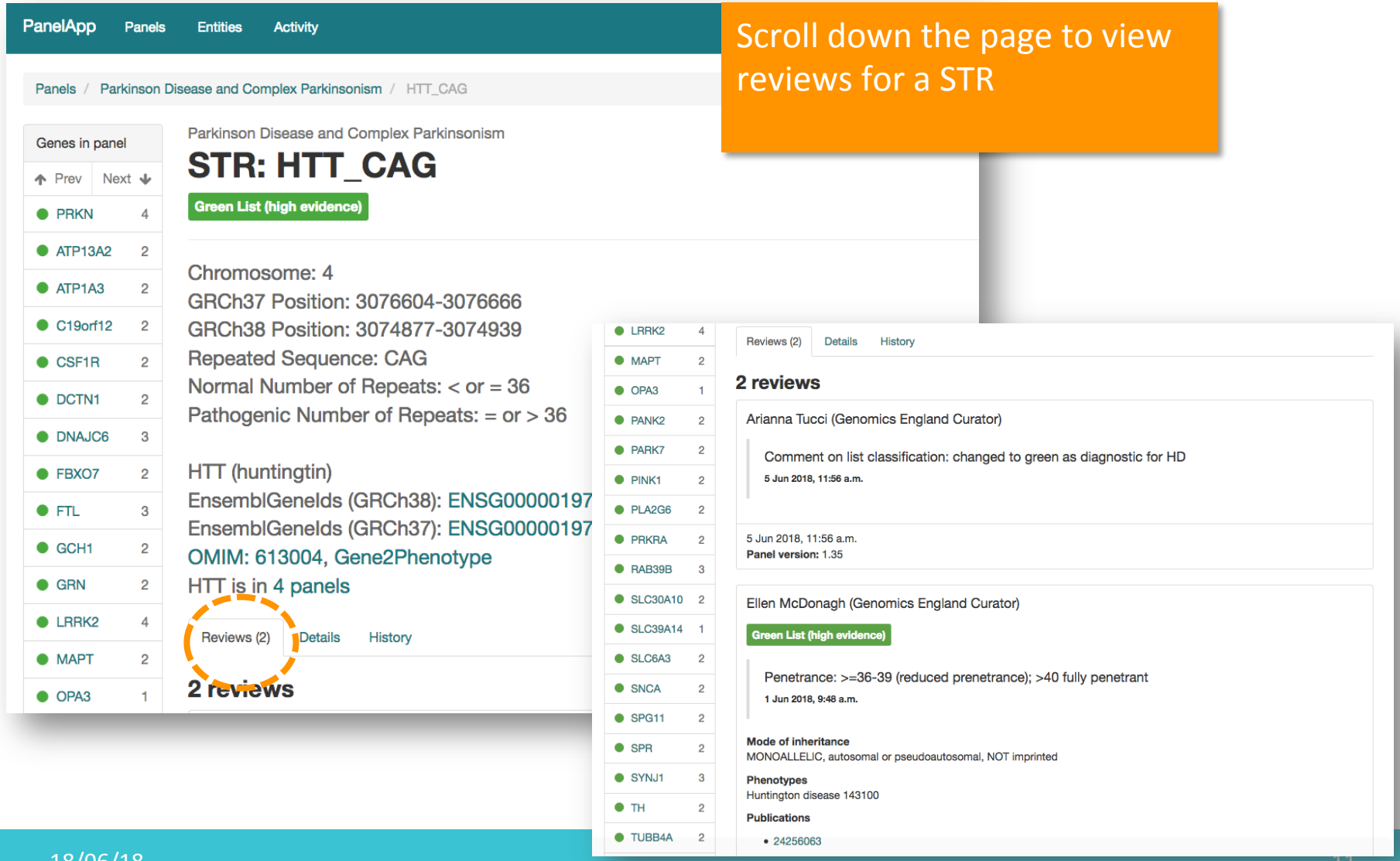
Find STRs on the associated gene page

Click on the STR to view it within a chosen panel



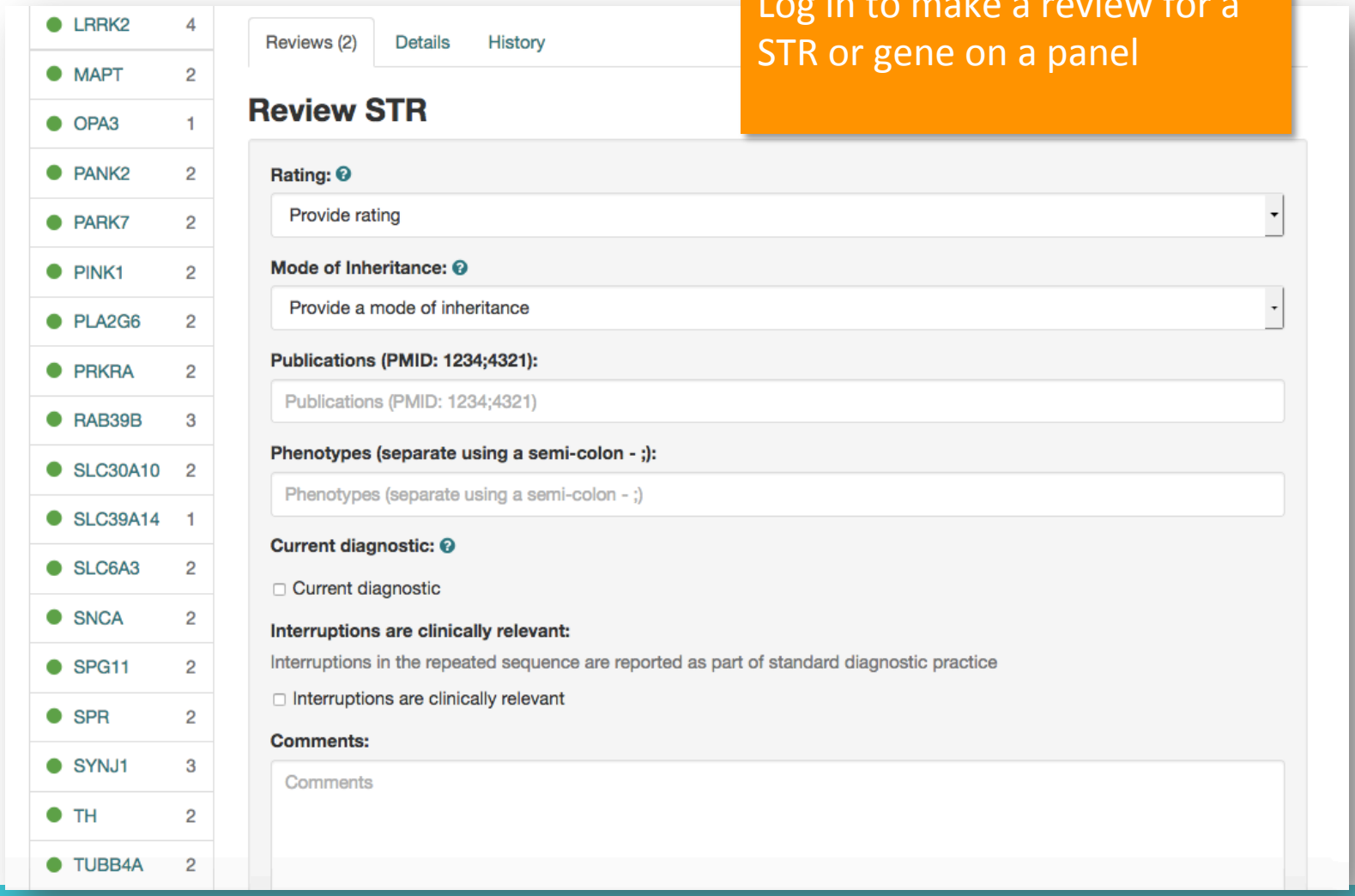
STR reviews

Scroll down the page to view reviews for a STR

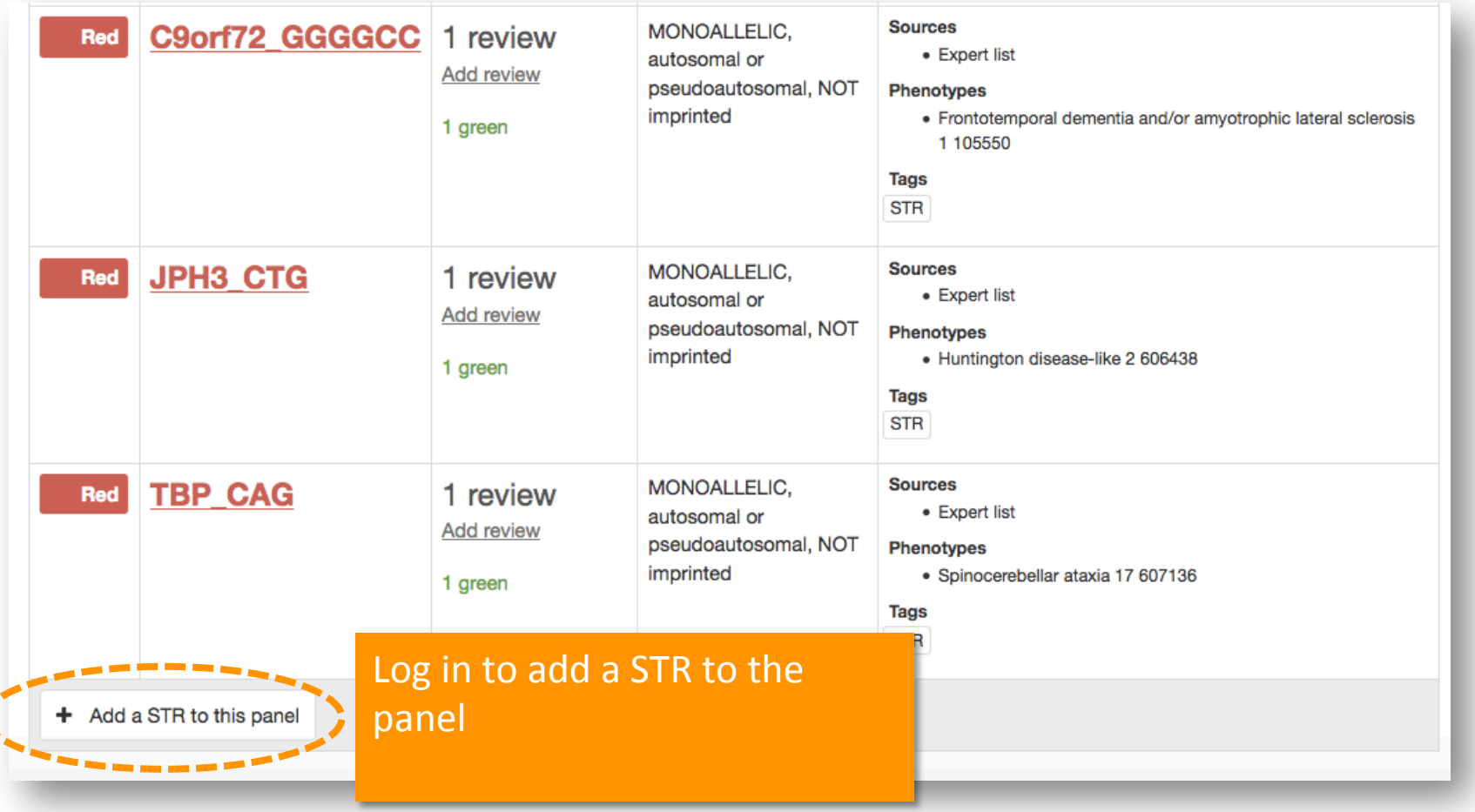


Submit a review

Log in to make a review for a STR or gene on a panel

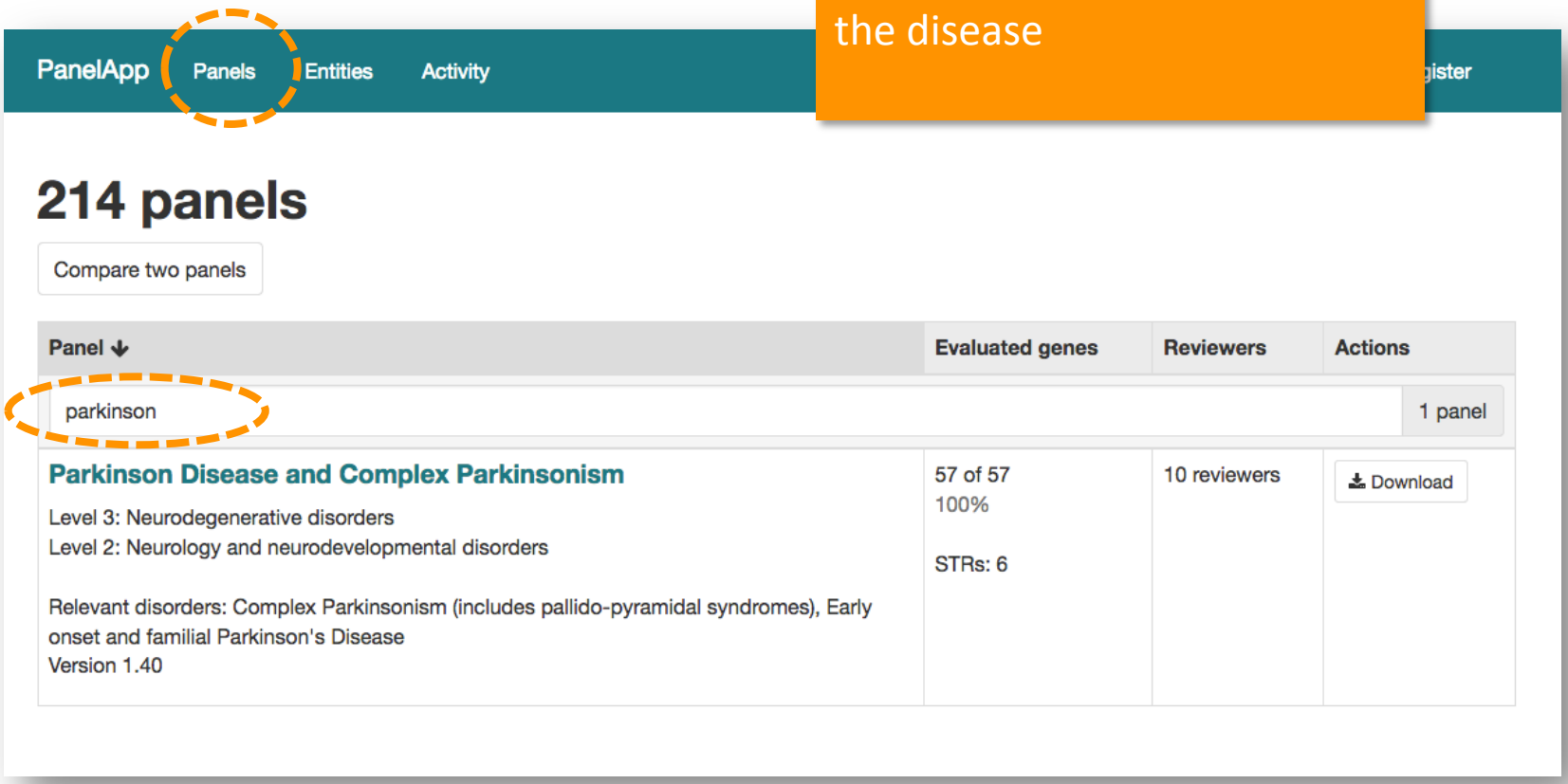


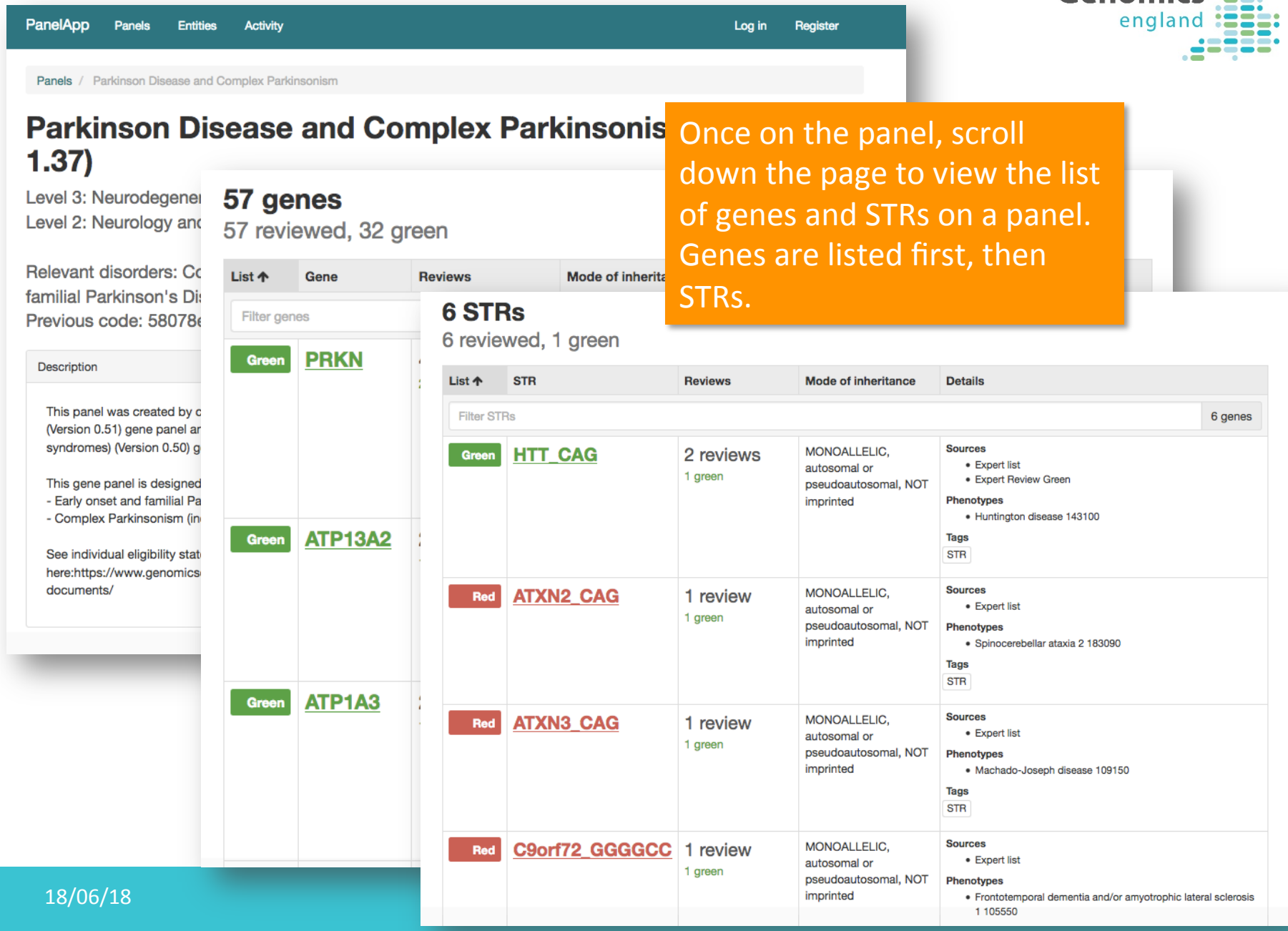
Add an STR to a panel as a reviewer



Find a panel

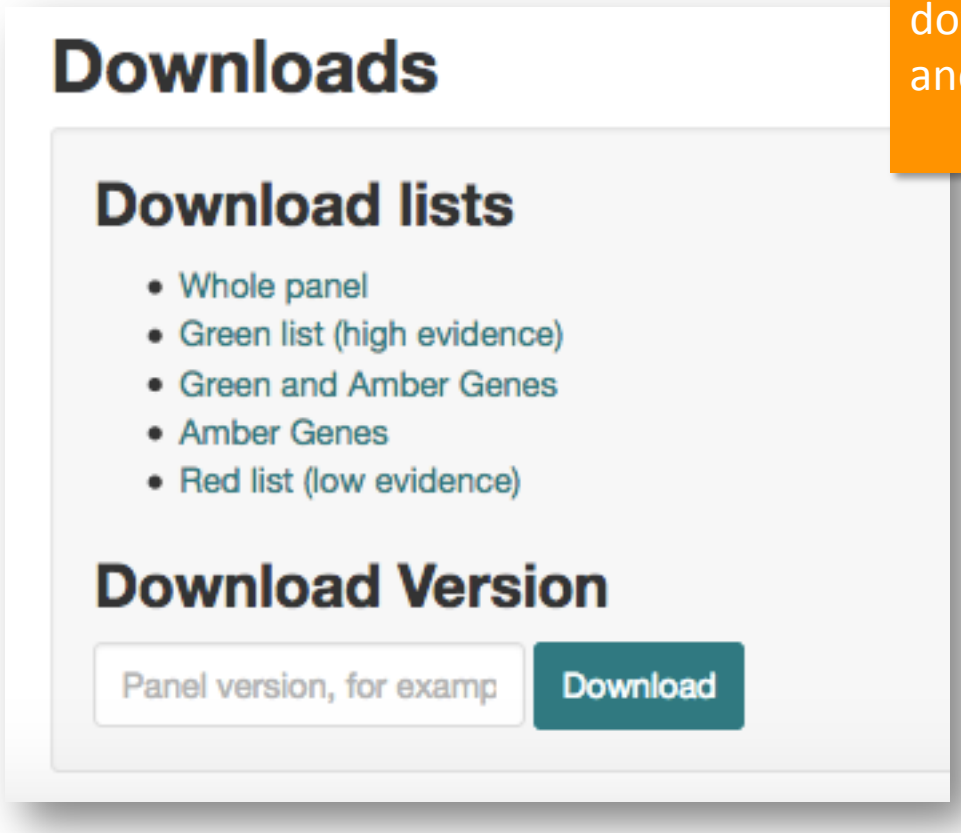
Go to the ‘Panels’ page and filter for key words related to the disease




PanelApp
Panels
Entities
Activity
Log in
Register
Panels / Parkinson Disease and Complex Parkinsonism
Parkinson Disease and Complex Parkinsonis
1.37)
Level 3: Neurodegener
Level 2: Neurology and
Relevant disorders: Co
familial Parkinson's Dis
Previous code: 58078e
Description
This panel was created by c
(Version 0.51) gene panel ar
syndromes) (Version 0.50) g
This gene panel is designed
- Early onset and familial Pa
- Complex Parkinsonism (in
See individual eligibility stat
here:https://www.genomics
documents/
57 genes
57 reviewed, 32 green
List
Gene
Reviews
Mode of inherita
Filter genes
Green
PRKN
Green
ATP13A2
Green
ATP1A3
Once on the panel, scroll down the page to view the list of genes and STRs on a panel. Genes are listed first, then STRs.
6 STRs
6 reviewed, 1 green
List
STR
Reviews
Mode of inheritance
Details
Filter STRs
6 genes
Green
HTT_CAG
2 reviews
1 green
MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Sources
Expert list
Expert Review Green
Phenotypes
Huntington disease 143100
Tags
STR
Red
ATXN2_CAG
1 review
1 green
MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Sources
Expert list
Phenotypes
Spinocerebellar ataxia 2 183090
Tags
STR
Red
ATXN3_CAG
1 review
1 green
MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Sources
Expert list
Phenotypes
Machado-Joseph disease 109150
Tags
STR
Red
C9orf72_GGGGCC
1 review
1 green
MONOALLELIC, autosomal or pseudoautosomal, NOT imprinted
Sources
Expert list
Phenotypes
Frontotemporal dementia and/or amyotrophic lateral sclerosis 1 105550

Downloads

Scroll down the page to download the panel – the download will include genes and STRs



Acknowledgements

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- **PanelApp Curators** (past & **present): Ellen McDonagh, Sarah Leigh, Rebecca Foulger, Louise Daugherty, Olivia Niblock, Eleanor Williams, Arianna Tucci, Helen Brittain, Rachel Jones**, Eik Haraldsdottir, Alice Gardham, Ellen Thomas, Richard Scott, Caroline Wright, Emma Baple, Damian Smedley, Chris Boustred, Kirsty McCaffrey, Chris Campbell.
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Any questions?

panelapp@genomicsengland.co.uk

@PanelAppTeam

#PanelApp

Find out more

https://
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