

RFC45: Gene panel information in Patient View

Title:	Gene panel information in Patient View
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RFC Type:	Exploratory
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1. What central problem does this RFC address?

Gene panel information is missing in patient view.

2. What feedback are you seeking from the Community?

Review of the proposed UI solution and information regarding similar efforts in other groups.

3. What is your proposed solution?

Add gene panel information to the patient view. Add it next to the genome overview tracks and in the mutations and CNA lists. See mockups proposed in this document.

4. How is the proposed RFC useful for the public cBioPortal?

Gene panel information has become an integral part of cBioPortal. Correctly reflecting this data across the different views is something all will benefit from.

5. How is the proposed RFC useful for local cBioPortal instance, e.g. at academic centers or pharmaceuticals?

Same as #4.

6. What is the impact of this RFC on the current refactoring efforts?

Refactoring of the Patient View is done, so this should not lead to any conflicts.

7. How will the work proposed within your RFC be maintained over the long-term?

No specific plans for now. Assuming this will become essential core functionality.

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Introduction

The Patient View is a great resource for visualizing serial samples; but as different samples are potentially profiled by different panels, support for gene panel information becomes key for this page.

For example, a patient has a tissue sample profiled at Panel X (300 gene panel) and then some serial plasma samples profiled at Panel Y (on a 50 gene panel). The Patient View is great for reviewing concordance in mutation status between these multiple samples but it's important for the user to bear in mind that not all the genes have been profiled on both gene panels. Currently this is not clear to casual users, and they could come away with a false impression of the data.

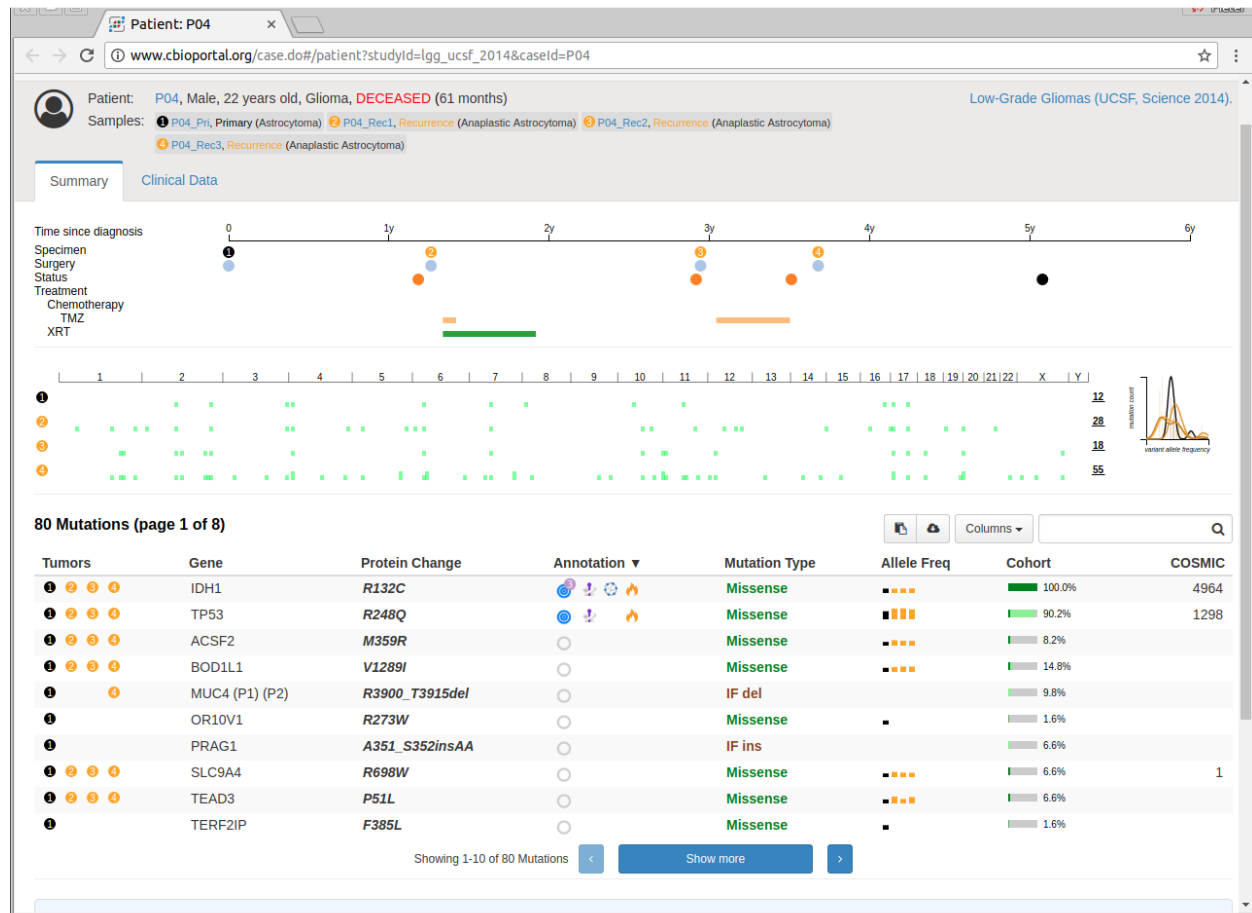


Figure: current patient view, showing multiple samples. In the current view, it is unclear which samples were profiled by which panel. It is also not clear which genes are covered by which panel. For example: why are there no PRAG1 mutations in samples 2, 3 and 4? Is this because it was not profiled in these samples, or is it because it is simply not found to be mutated in these samples?

Proposed solution

Panel information

The proposed solution is to add gene panel information to the patient view, in two places:

1. Next to the genome overview tracks
2. In "Tumor" column in the mutations and CNA tables in the patient view.

See mockups below.

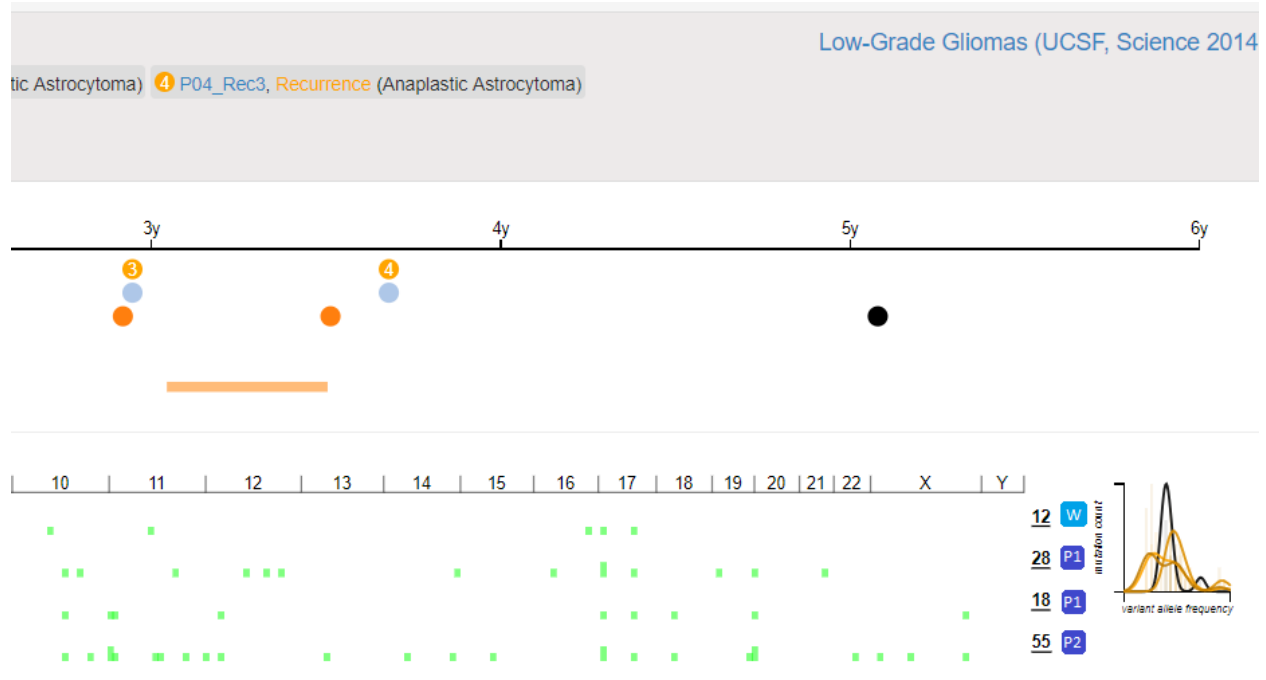


Figure 1: gene panel icons added next to the sample tracks, to inform the user about which panel was used to profile each sample. When gene panel information is not given for the sample, cBioPortal will assume it has been profiled as Whole Exome sequencing. This is shown by the  icon in the first sample.

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Tumors	Gene	Protein Change	Annotation ▼
1 2 3 4	IDH1	R132C	   
1 2 3 4	TP53	R248Q	   
1 2 3 4	ACSF2	M359R	
1 2 3 —	BOD1L1	V1289I	
1 — 4	MUC4	R3900_T3915del	
1	OR10V1	R273W	
1	PRAG1	A351_S352insAA	
1 2 3 4	SLC9A4	R698W	
1 2 3 4	TEAD3	P51L	
1 — — —	TERF2IP	F385L	

Figure 2: a new “Not profiled” icon will be added to the tumors column. This icon should appear instead of the sample icon whenever the gene displayed in the table row is known to **not** have been profiled in that sample. E.g. in this screenshot example, BOD1L1 is not part of the panel used for sample 4, therefore sample 4 placeholder has a — icon indicating BOD1L1 has not been profiled in this sample.

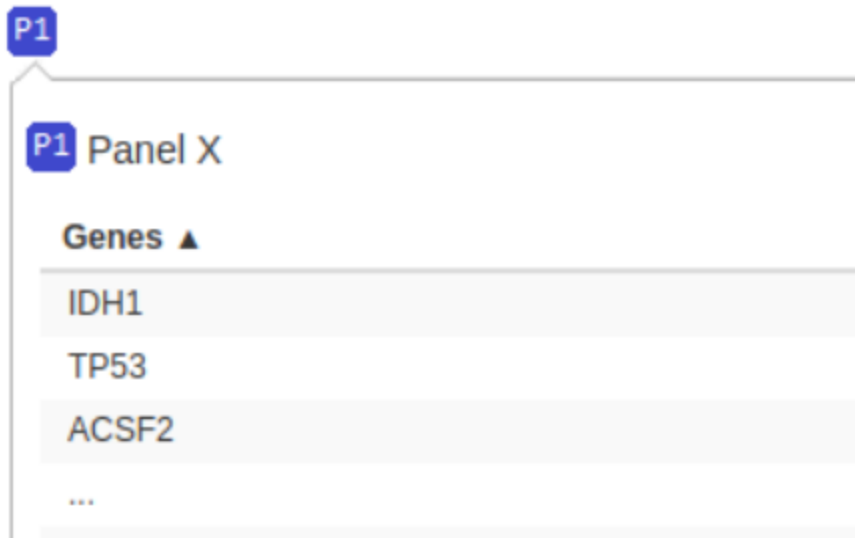


Figure 3: tooltip: hovering the panel icon should display the relevant panel information + the genes that are part of this panel.

Filtering

To enable filtering only the mutations that are found in genes profiled by all panels, a filter button should be added for the “Genes” column. When clicked, it should display the filter options for the “Gene” column, namely: (1) filter genes profiled in any sample, (2) filter only genes profiled in **all** samples. See figure below.

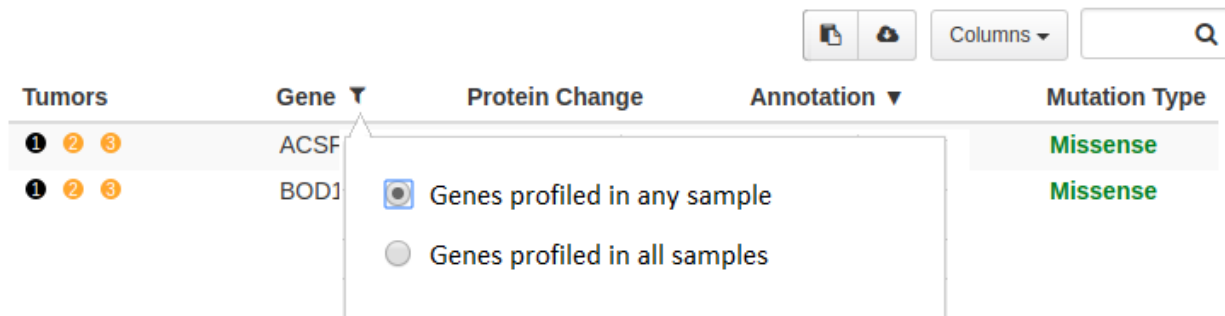


Figure 4: new filter button for Gene column. Show here for mutations table, but **also** to be added for CNA table.

Default filters

By default, the Gene column filter will be to have “Genes profiled in **any** sample”. However, it should be possible to configure a **portal.properties** property to have “Genes profiled in **all** samples” as the default filter on startup of the Patient View. This single property will apply to both mutations and CNA tables.

Updating the genome overview

Currently there is no link between what is filtered in the mutations table and the genome mutational burden overview on the top part of the Patient View. To better compare samples profiled in different panels, it would be nice if the genome overview would reflect the filtering done in the table.



Figure 5: (alternative to Figure 6below) only the filtered mutations are shown in the genome overview. Shown here for mutation data, but **also** to be done for CNA tracks.



Figure 6: (alternative to Figure 5) the filtered mutations are highlighted in the genome overview. Shown here for mutation data, but **also** to be done for CNA tracks.

When updating the genome overview, it is also important to update the related histogram on the right side. The panel icons should also appear in the histogram view as well.

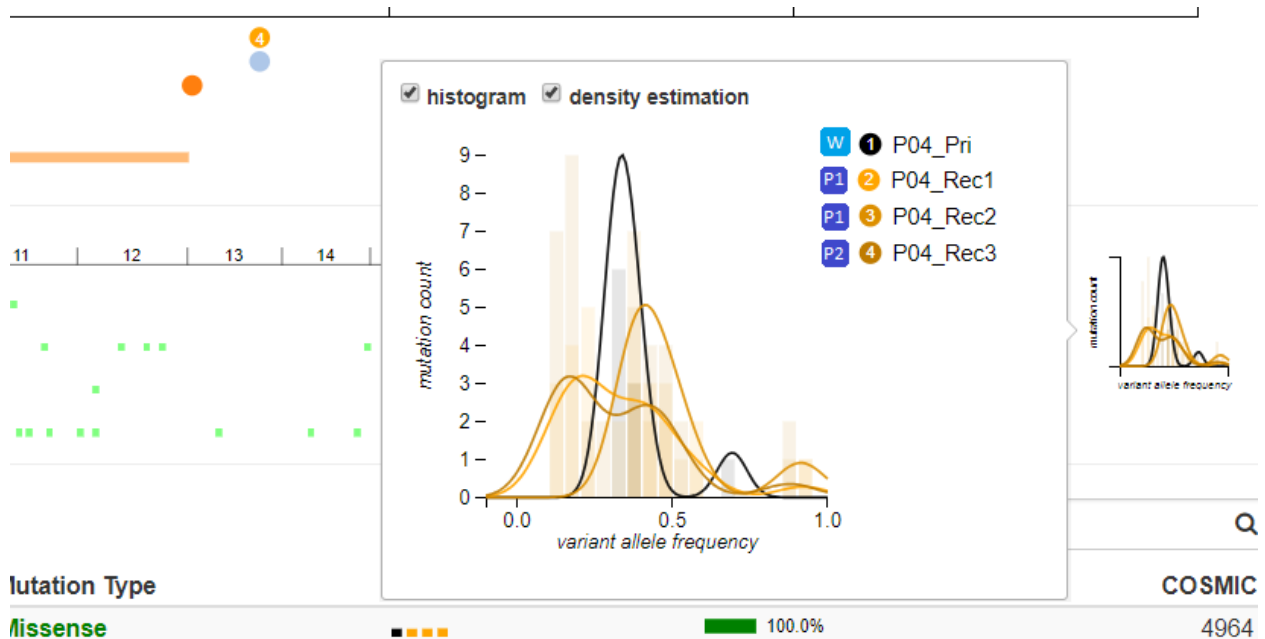


Figure 7: mutations histogram view to be automatically updated when filtering of the table changes.

Renaming and resizing the tumors column

To keep the naming consistent with other parts of the portal, the “Tumors” column will be renamed to “Samples”. Also, it should be clear to the user that the “Samples” column is resizable (in some studies there might be up to 20 samples per patient). See figure below.

Samples	Gene	Protein
1 2 3 4	IDH1	R132
1 2 3 4	TP53	R248

Figure 8: Tumors column now called “Samples”. There is a line next to it to indicate it is resizable.

Samples	Gene	Protein
1 2 3 4	IDH1	R132
1 2 3 4	TP53	R248

Figure 9: when resize line is hovered over by mouse, the icon changes to a resize icon and the user can resize the Samples column.

Downloading the data

The Panel information should be added to the downloaded TSV. Example:

	A	B	C	D	
1	Tumors	Gene	Panel	Protein Change	Annotation
2	P04_Pri	IDH1	WES	R132C	OncoKB: Oncoge
3	P04_Rec1	IDH1	Panel X	R132C	OncoKB: Oncoge
4	P04_Rec2	IDH1	Panel X	R132C	OncoKB: Oncoge
5	P04_Rec3	IDH1	Panel Y	R132C	OncoKB: Oncoge
6	P04_Pri	TP53	WES	R248Q	OncoKB: Likely C
7	P04_Rec1	TP53	Panel X	R248Q	OncoKB: Likely C
8	P04_Rec2	TP53	Panel X	R248Q	OncoKB: Likely C
9	P04_Rec3	TP53	Panel Y	R248Q	OncoKB: Likely C

Figure 10: New "Panel" column added to the downloaded data.