CONTROLS PANEL (LEFT)

SUBPANEL "VARIANT CALL SET"

Contains drop-down to select call set

Comment [PV1]: Proposal: two drop-downs: "Sample set" and "Calling method"

SUBPANEL "VARIANT FILTERS"

Contains filters acting on a variant position (i.e. over all samples)

GATK CALLSETS

- Variant type: [x] SNP [x] INDEL
- SNP min VQSLOD: --|--
- INDEL min VQSLOD: --|--
- Min. % called: --|-- (100% by default)
- [x] Parent calls not missing
- [x] Segregating (selected by default, means non-segregating variants - where all calls are ALT - are hidden)
- One checkbox per FILTER (all selected by default, selected means FILTER is applied, i.e., only PASS variants are shown by default)

CORTEX CALLSETS

- Variant type: [x] SNP [x] INDEL
- Min. SITE_CONF: --|-- (50 by default)
- Min. % called: --|-- (100% by default)
- [x] Parent calls not missing
- [x] Segregating
- One checkbox per FILTER (all selected by default, selected means FILTER is applied, i.e., only PASS variants are shown by default)

Comment [PV2]: Do we want to display the filter names as set in the VCF files? Or do we want to replace this with more human readable names?

SUBPANEL "CALL FILTERS"

Contains filters acting on individual calls (i.e. a specific variant called on a specific sample)

GATK CALLSETS

- Min. DP: --|-- (5 by default)
- Min. GQ: --|-- (99 by default)

CORTEX CALLSETS

- Min. GT_CONF: --|-- (50 by default)

SUBPANEL "DISPLAY SETTINGS"

- Color by: inheritance | ref/non-ref (radio button group, inheritance selected by default)
- [x] Equidistant blocks (selected by default)
- [x] Allow small blocks (selected by default)
- [x] Show magnifying glass (not selected by default)
- [Sort by parents] button

Comment [PV3]: If this should be selected by default, I think there is little reason to have this option in there at all.

Comment [PV4]: This was some kind of toy experiment; I propose not to leave it as it adds little value.

GENOTYPE BROWSER PANEL

MISCELLANEOUS

- Parents at top.
- Sample names: separate the different sample identifiers using "/" rather than "__" and make enough space.
- Move "find feature" to top (next to chromosome selector and navigation controls) and rename as "find gene"
- Remove allele depths area
- Change color of find gene results
- Add pseudogenes to gene annotations

Comment [PV5]: How should this behave in association with the "Sort by parents" button? Currently, this button puts the parents at top & bottom, and sorts all the progeny according to the distance to the parents

Comment [PV6]: Would it make sense to show only one identifier type at a time, and let the user chose which one from the "Display settings"? This would allow us to save some space

Comment [PV7]: I assume this refers to the red/blue colors vertical bar that appears on the right if you hover over a SNP?

GENOME TRACKS

- Add nucleotide sequence track
- Add genome region classification track:
  - Core: grey
  - SubtelomericHypervariable: red
  - InternalHypervariable: red
  - SubtelomericRepeat: orange
  - Centromere: black
- Remove average purity track
- Remove average coverage track

- If variant type track is fixed at bottom, should be properly fixed, and scroll bar should be within the genotype pane (so obvious if there are more samples offscreen)

COLORING OF CALL BLOCKS

- Color blocks red/blue not pink/blue, no variable hue
- Color by inheritance by default
- In color by inheritance, use green for parents both ALT, orange for parents both REF
- Don't show variable block heights
- White (with dot-like?) if missing call, grey block if filtered call

Comment [PV8]: And black for Mendelian error?

HOVER ACTIONS

When hover over call block, in fixed bar at bottom of panel, show e.g:

```
Variant: REF:A ALT:T FILTER:PASS   Call: GT:0 GQ:99 DP:14 AD:12,4 PL:123,0
(i.e., whatever FORMAT fields are available)
```

Comment [PV9]: Note: showing all call details in a hover action requires all information to be pre-emptively fetched, which will result in a drastic speed reduction.
Proposal: only show those that we need for filtering (DP,AD,GQ,GT_CONF), and show the rest in the popup.

CLICK ACTIONS

When click on call block, get popup with:

- Call details: `GT:0 GQ:99 DP:14 AD:12,4 PL:123,0`
- Variant details: fixed fields from VCF (CHROM, POS, ..., all INFO fields)
- [genome accessibility] button to navigate to genome accessibility page centred on variant