**Filtering strategies workshop: question sheet**

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| **Q1** | From the ANNOVAR log file, how many refGene transcripts were used for annotation? |
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| **Q2** | Do the percentages of shared variants agree with reported pedigree? |
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| **Q3** | Is there any evidence to suggest that the parents are related? |
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| **Q4** | Compare the proportion variants shared between unrelated individuals with the result from example 1 and give a reason which accounts for most of the difference. |
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| **Q5** | How many variants are identified in each type of Mendelian error? |
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| **Q6** | How does the number of de-novo mutations compare with expectation? |
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| **Q7** | How many plausible de-novo variants meet the search criteria? |
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| **Q8** | Which variant is most likely to cause the disease in the child? |
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| **Q9** | What proportion of reads have the mutant allele (mutant allele frequency)? |
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| **Q10** | Is there anything unusual about the mutant allele frequency and if so how could it be related to the disease? |
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| **Q11** | Does either chromosome have any evidence for aUPD and if so what are the approximate genomic coordinates in megabses? |
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| **Q12** | Which variant is most likely to cause the clonal proliferation of erythrocytes (polycythemia vera) in this patient? |
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