

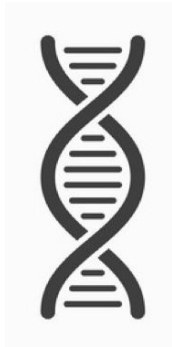
Visualizing Structural Genomic Variants



CPSC547 Project Pitch
Janet Li & Armita Safa

Genome Sequencing

- Figuring out the order of DNA nucleotides



Sequencing

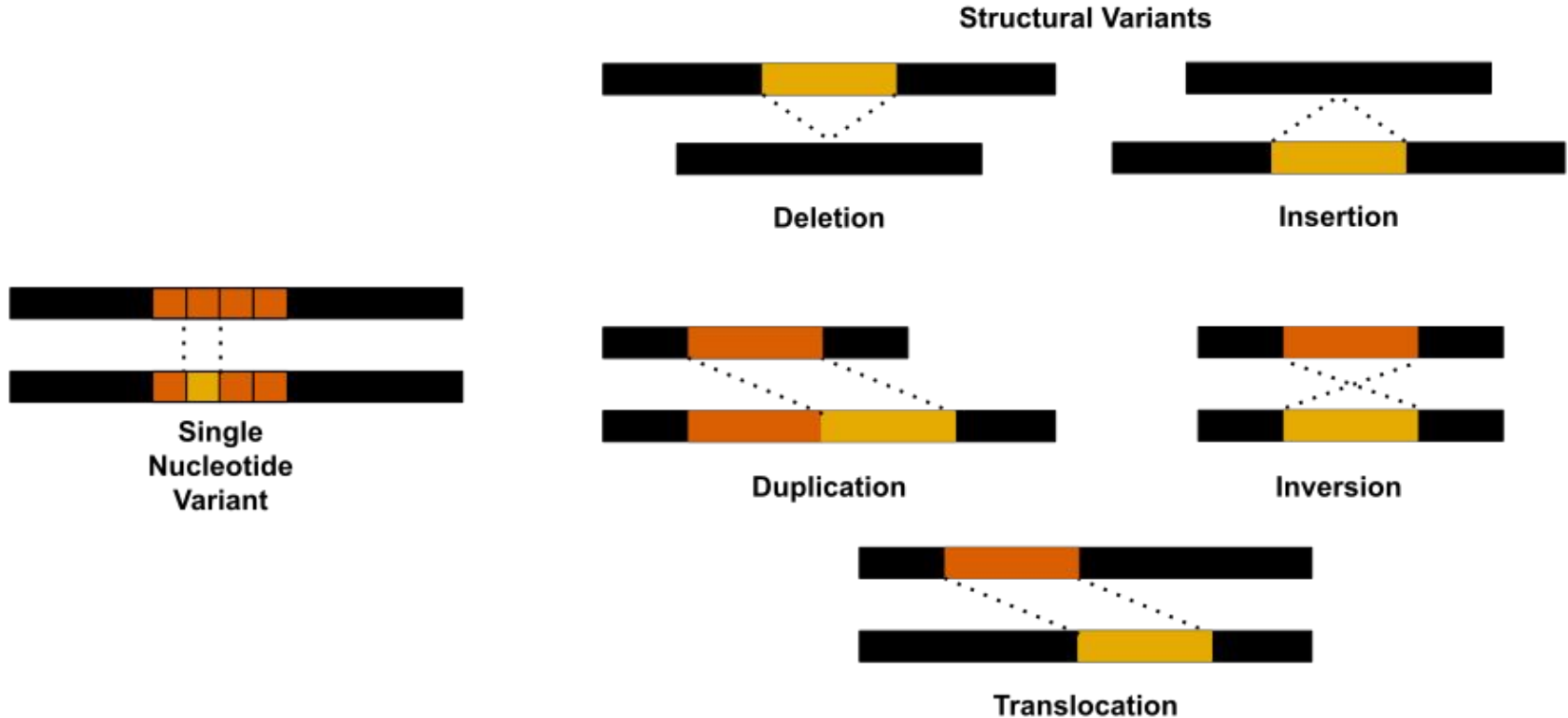


ACGCTGCAT...

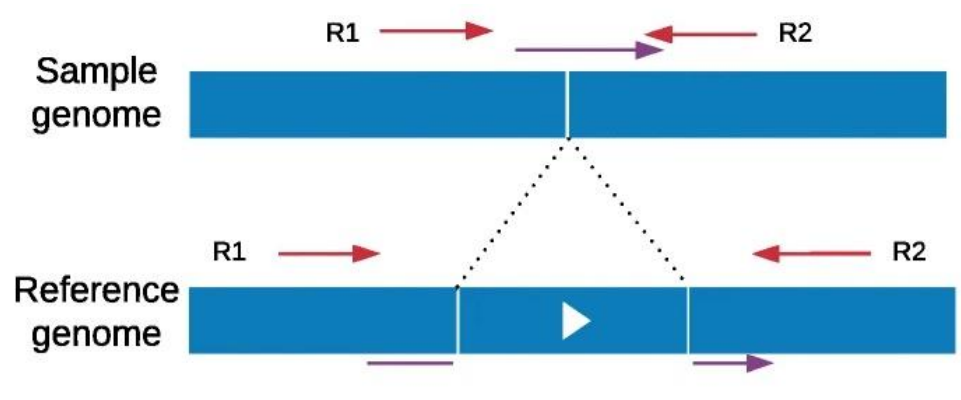
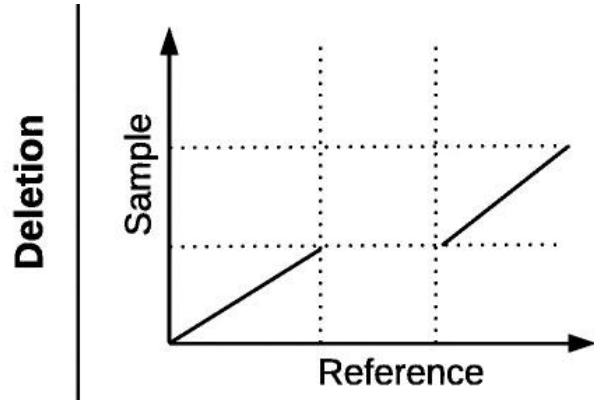
Genomic Variants

- Defined as alterations in the genome
- Impact evolutionary processes [Weischenfeldt et al, 2013]
- Traits and appearance – hair color, blood type, etc
- Susceptibility to diseases
 - Genomic variants have been associated with cognitive disabilities, predispositions to obesity, cancer and other maladies [Sudmant et al., 2015]
 - Large recurrent microdeletions associated with schizophrenia [Stefansson *et al.*, 2008]
 - The role of rare structural variants in the genetics of autism spectrum disorders [Kusenda *et al.*, 2009]

Genomic Variants



SV Detection



Example: Circos plots

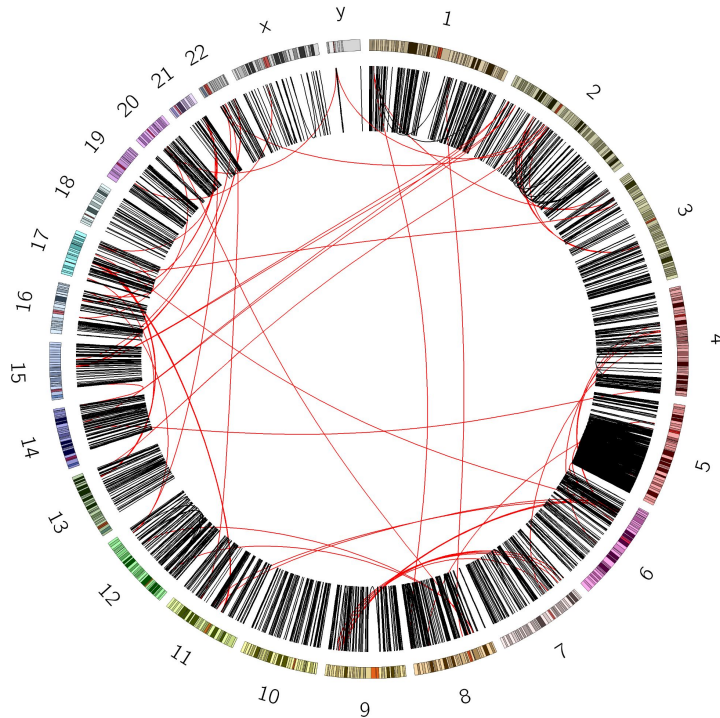
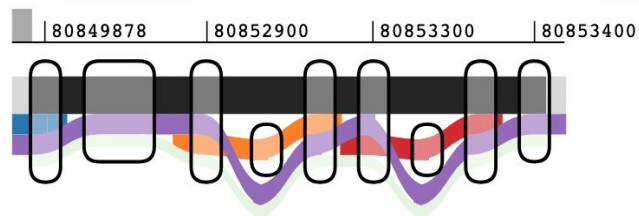


Image: <https://training.galaxyproject.org/training-material/topics/visualisation/tutorials/circos/tutorial.html>
Kryzwiniski et al. (2009). Genome Res. 19(9). doi: 10.1101/gr.092759.109

Example: Graph-based view

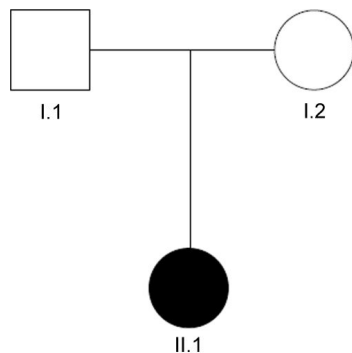
Graph View: SequenceTubeMap



Color	Trackname	Show Track
■	chr12	<input checked="" type="checkbox"/>
■	ins_chr12:80849878&chr12:80849878_NONE	<input checked="" type="checkbox"/>
■	ins_chr12:80853221&chr12:80853221_NONE	<input checked="" type="checkbox"/>
■	ins_chr12:80853303&chr12:80853303_NONE	<input checked="" type="checkbox"/>
■	inv_chr12:80842171&chr12:80861781	<input checked="" type="checkbox"/>

Visualizing Multiple SVs

- It's often interesting to compare genomic differences between individuals
- Trio analysis (parents + child)
 - Was an SV inherited, or did it occur as a *de novo* alteration?
- Tumour/normal comparisons
 - Which SVs are present in tumour tissue and may be driving more mutations?



Project

- View and compare SVs in multiple genomes
- Show positions of genes related to a certain disease
- Potential dataset sources:
 - Genome in a Bottle (variant calls)
 - 1000 Genomes Project (variant calls)
 - DisGeNET (disease-gene associations)