

Genomic Variants of K562 & HepG2

NEW RESOURCES FOR ENCODE

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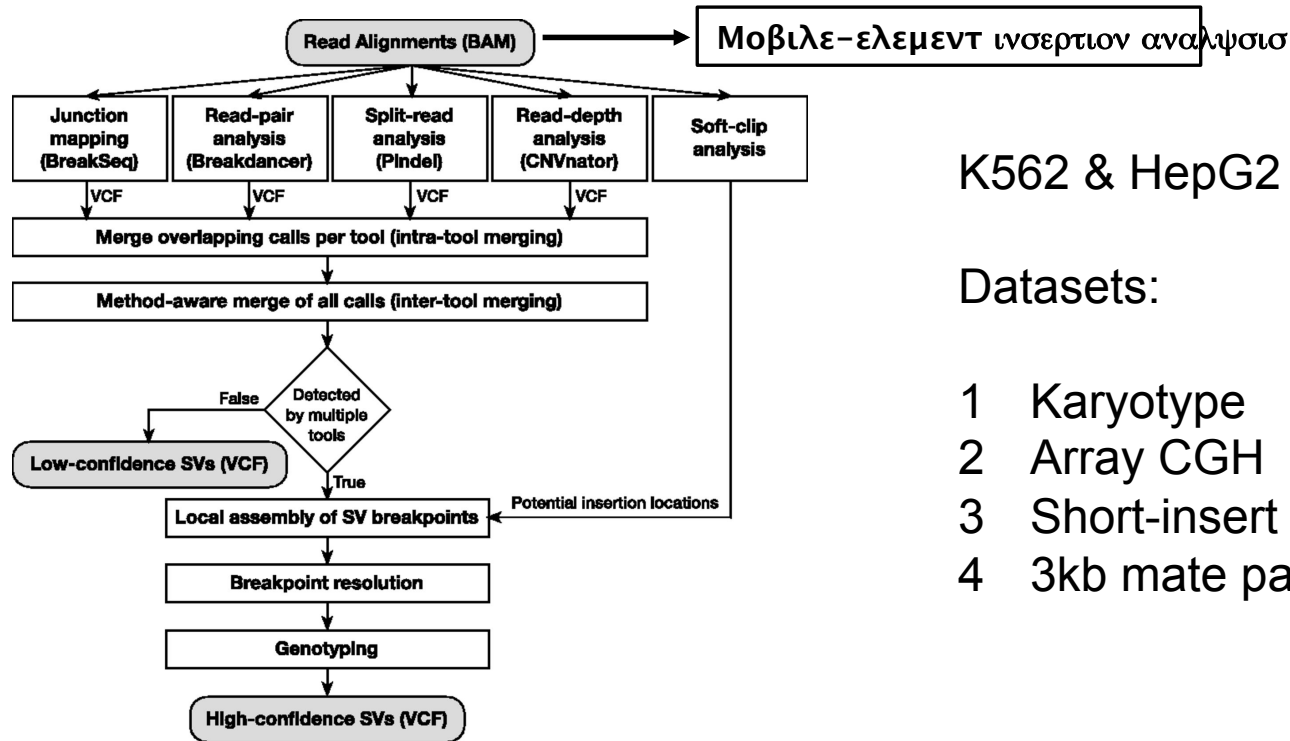
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High-level view of methodology



K562 & HepG2

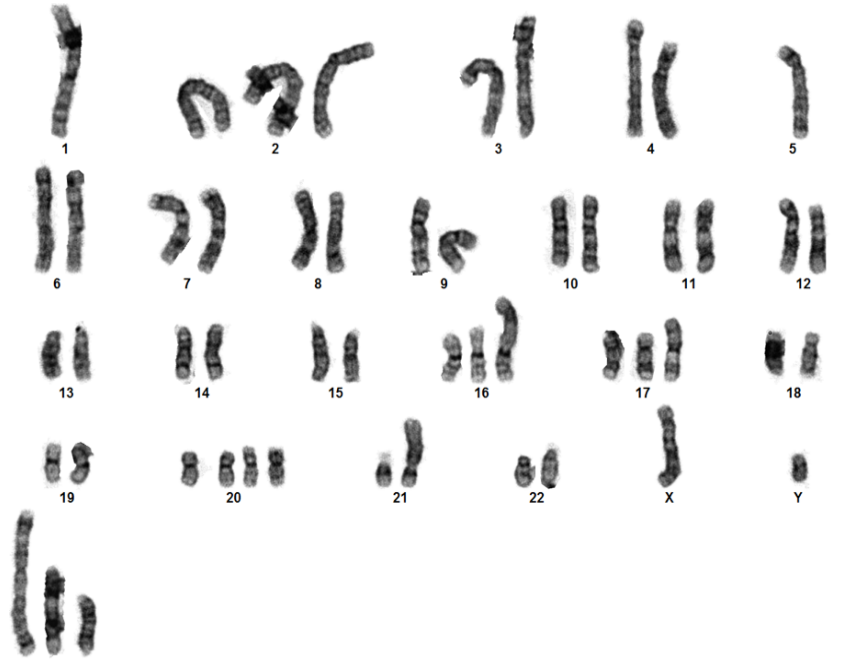
Datasets:

- 1 Karyotype
- 2 Array CGH
- 3 Short-insert WGS (60-70X)
- 4 3kb mate pair WGS (10X)

Karyotype

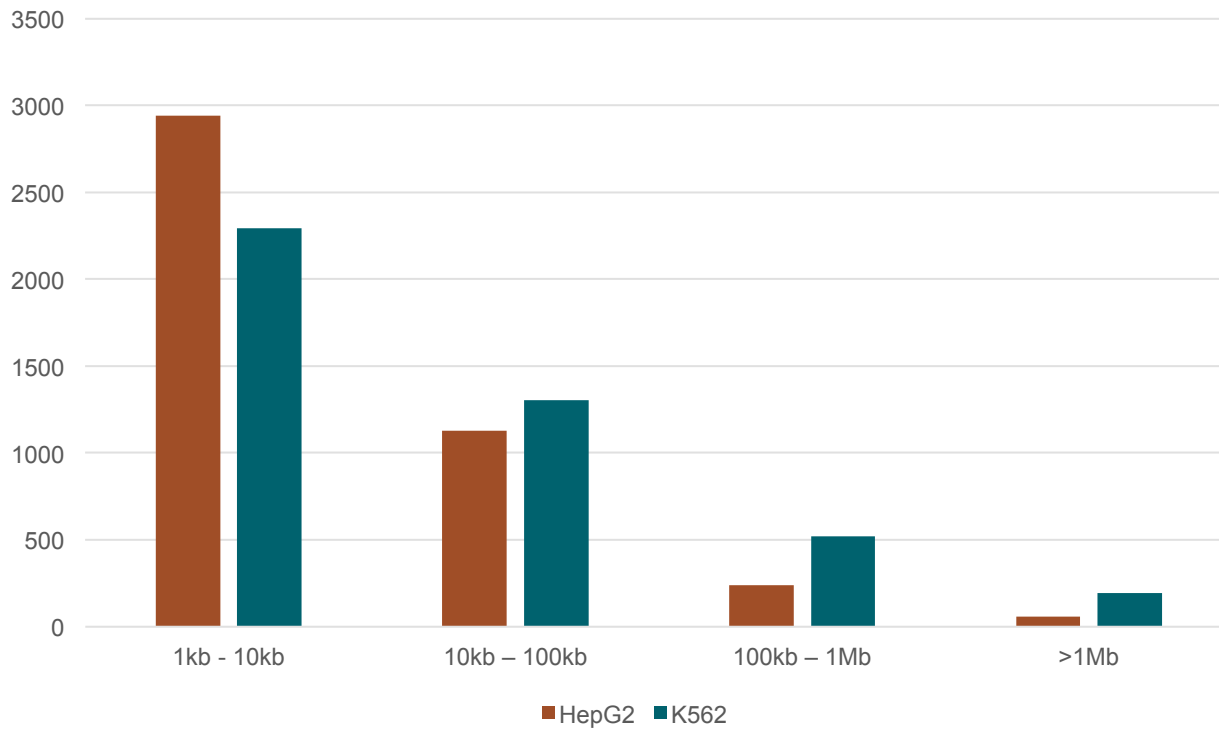


K562



HepG2

Medium and Large CNVs



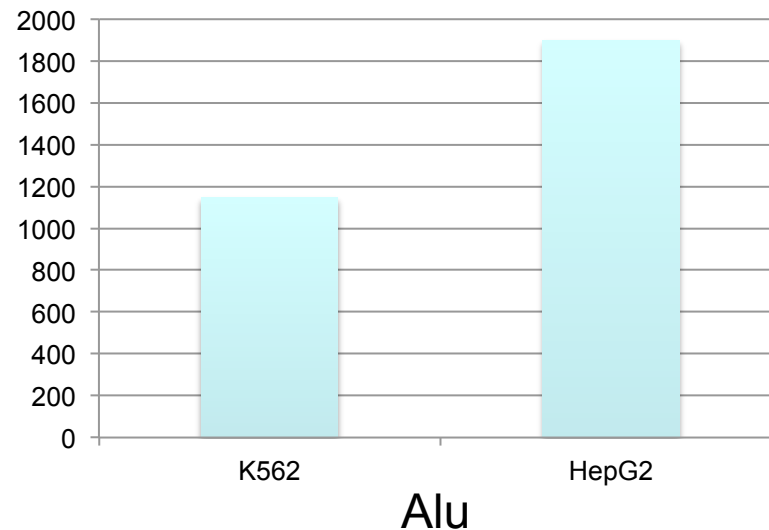
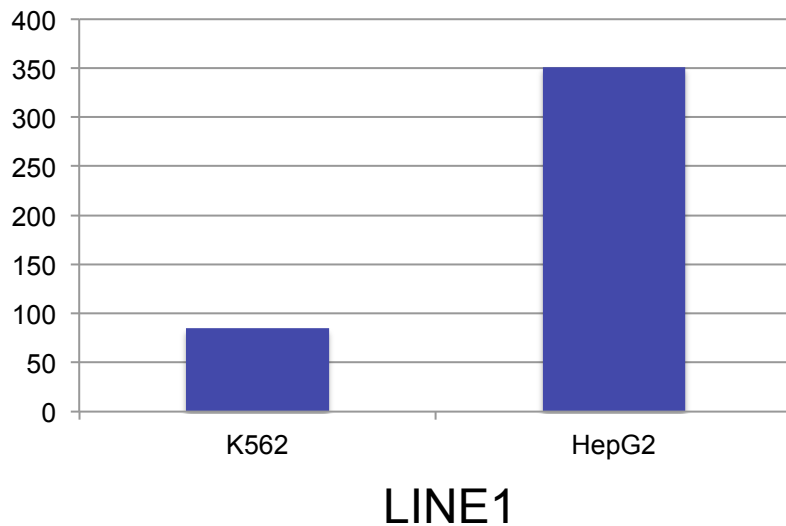
	HepG2	K562
SNPs	3.77 million	3.44 million
Homozygous SNPs	39%	49%
Heterozygous SNPs	61%	51%
Indels	~849,000	~679,000

Mobile Element Insertion Analysis

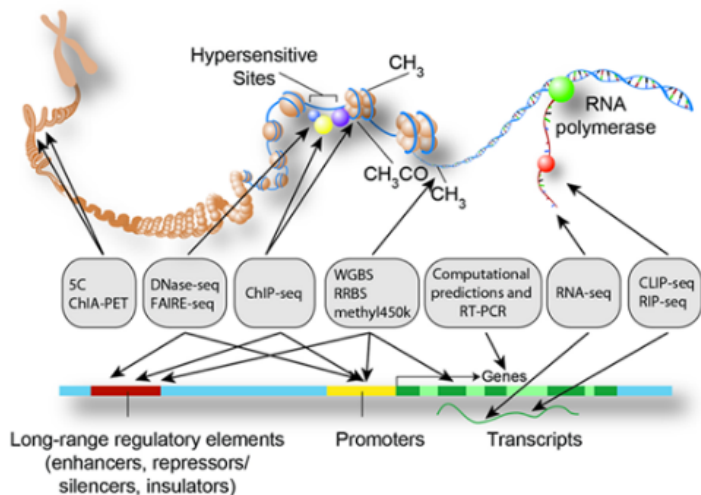
Xiaowei Zhu
Reenal
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K526		HepG2	
Alu	L1	Alu	L1
1147	85	1899	351

Secondary validation rate: ~95%
(PCR & Sanger sequencing)



ENCODE: Encyclopedia of DNA Elements



The ENCODE (Encyclopedia of DNA Elements) Consortium is an international collaboration of research groups funded by the National Human Genome Research Institute (NHGRI). The goal of ENCODE is to build a comprehensive parts list of functional elements in the human genome, including elements that act at the protein and RNA levels, and regulatory elements that control cells and circumstances in which a gene is active.

Image credits: Darryl Leja (NHGRI), Ian Dunham (EBI), Michael Pazin (NHGRI)