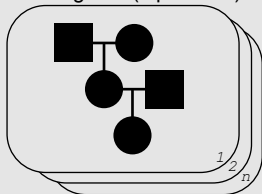


# Mutation Rate Estimation

## Calling Variants in a Set of Pedigrees

Sample of Extended Pedigrees (if possible)

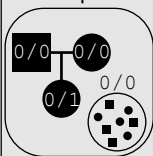


Candidate Mutations from Variant Calling

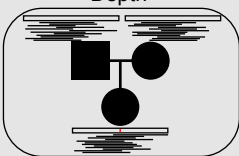
#	CHROM	POS	ID	REF	ALT
$r_1$	$s_1$	.	A	G	
$r_1$	$s_2$	.	C	T	
$r_1$	$s_3$	.	A	G	
$r_2$	$s_4$	.	C	T	
.	.	.	.	.	.
.	.	.	.	.	.
$r_1$	$s_m$	.	C	T	

## Variant Filtering to Remove False Mutations

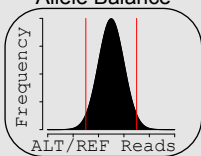
Unique



Depth



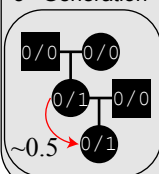
Allele Balance



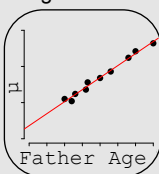
$$\mu = \frac{\text{Number of Mutations Passing Filters}}{\text{Number of Callable Sites} \times 2}$$

## Validation and Analysis

Transmission to 3<sup>rd</sup> Generation



Model Paternal Age Effects



Biases in the Mutation Spectrum

