

1



Sample
Collection

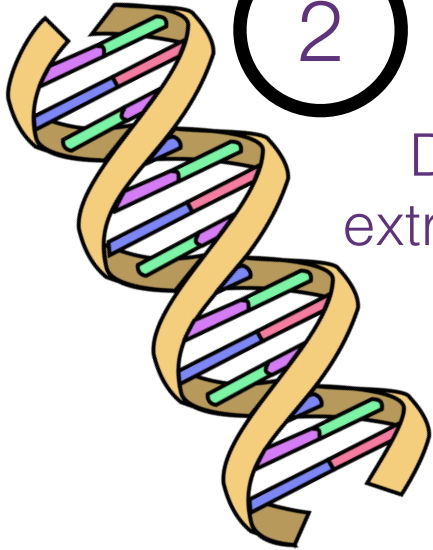
Sample
swap!

Sample
contamination

5

Patient receives
genetic diagnosis

2



DNA
extraction

3

Sequence
exonic
regions of
the genome



Probes fail
and too few
regions are
captured

4

Computational
analysis and
variant calling

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	57762	57770	57778	57794	57802
1	2234791	.	C	T	6061	PASS	.	GT:DP:GQ:PL	0/0:19:76:0,57,255	0/0:36			
1	12004626	.	C	T	2084	PASS	.	GT:DP:GQ:PL	0/0:279:99:0,255,255				
1	12004663	.	G	A	1463	PASS	.	GT:DP:GQ:PL	0/0:279:99:0,255,255				
1	12004675	.	C	T	2708	PASS	.	GT:DP:GQ:PL	0/0:279:99:0,255,255				
1	12004682	.	T	C	419	PASS	.	GT:DP:GQ:PL	0/0:173:99:0,255,255				
1	12004694	.	C	T	196	PASS	.	GT:DP:GQ:PL	0/0:173:99:0,255,255				
1	12009845	.	G	A	197	PASS	.	GT:DP:GQ:PL	0/0:22:99:0,66,255				
1	12009861	.	A	G	196	PASS	.	GT:DP:GQ:PL	0/0:22:99:0,66,255				
1	12009907	.	G	A	92	PASS	.	GT:DP:GQ:PL	0/0:43:99:0,129,255				

Celebrate a successful experiment!
Cool moves code

Bug in your
computer
code

Technology changes quickly!
Speed code