

# *CloudMap: A Cloud-based Pipeline for Analysis of Mutant Genome Sequences*

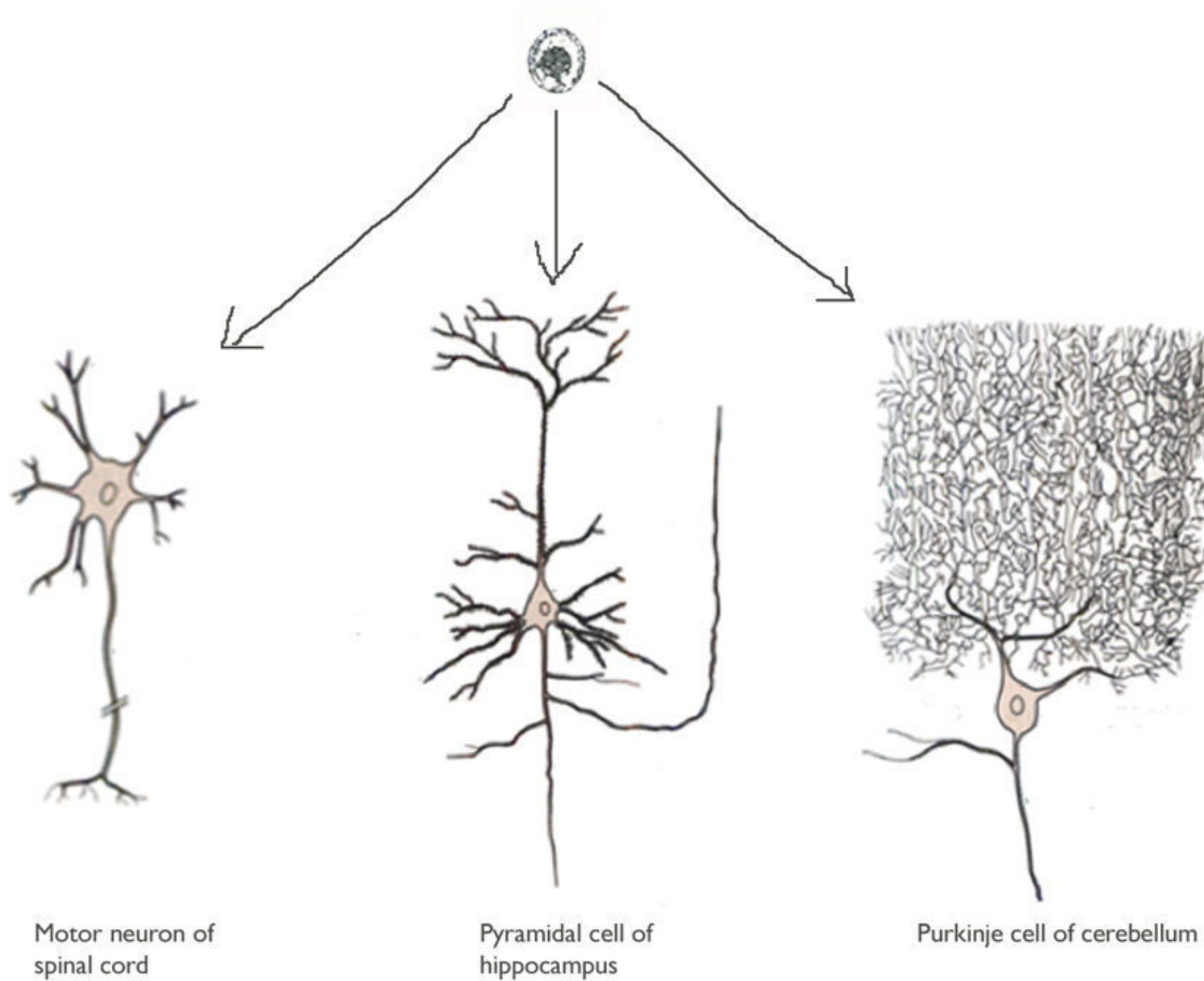
<https://test.g2.bx.psu.edu/u/gal40/p/cloudmap>

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Columbia University

<sup>2</sup> Center for Comparative Genomics and Bioinformatics,  
Penn State University

# *Identifying molecular mechanisms that create neuronal diversity*



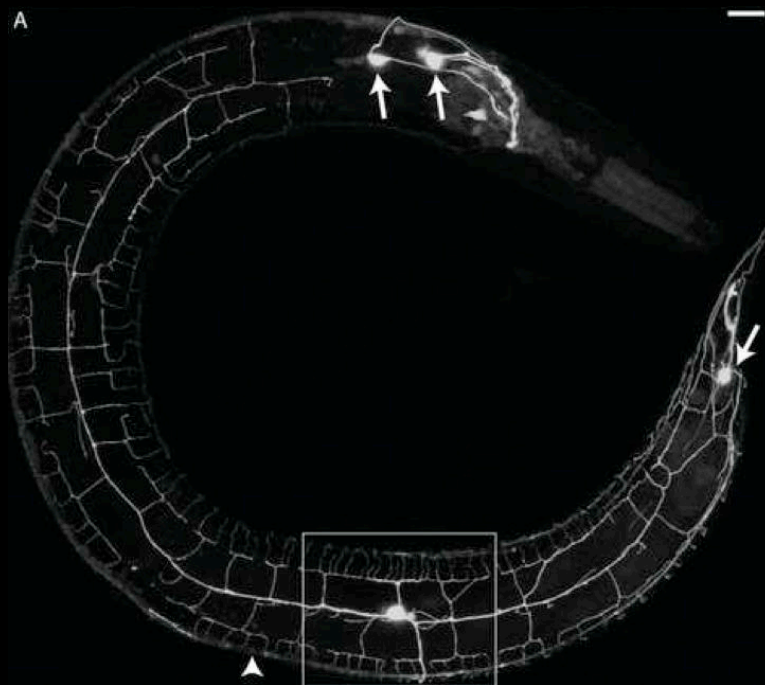
adapted from  
Kandel, 2000

# *C. elegans*: an ideal model organism



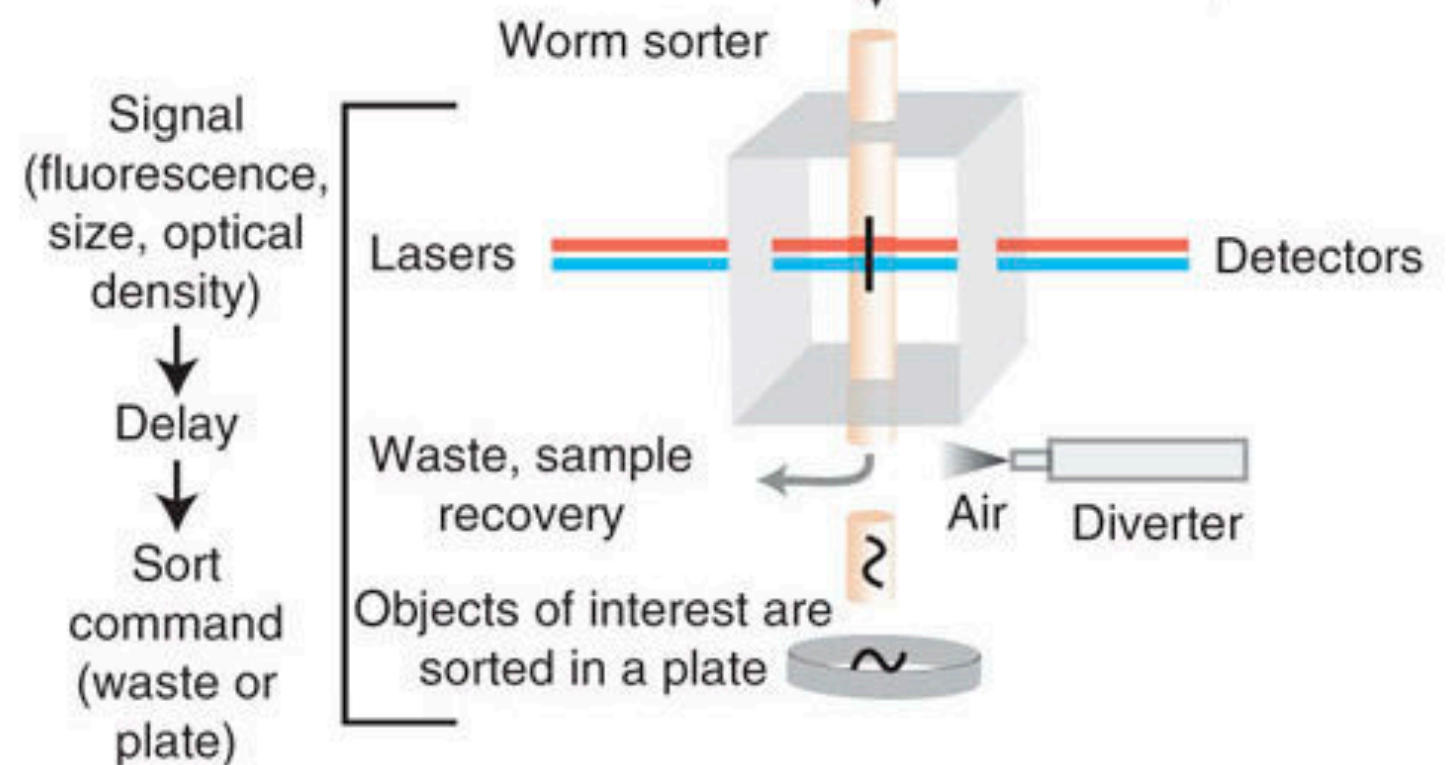
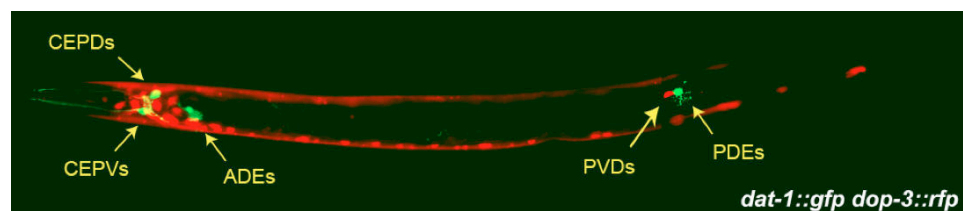
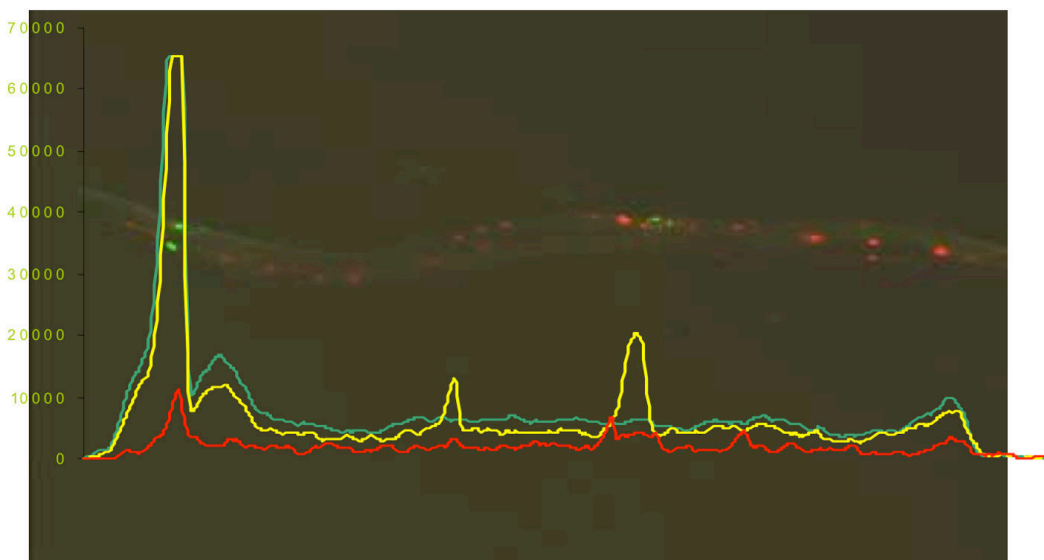
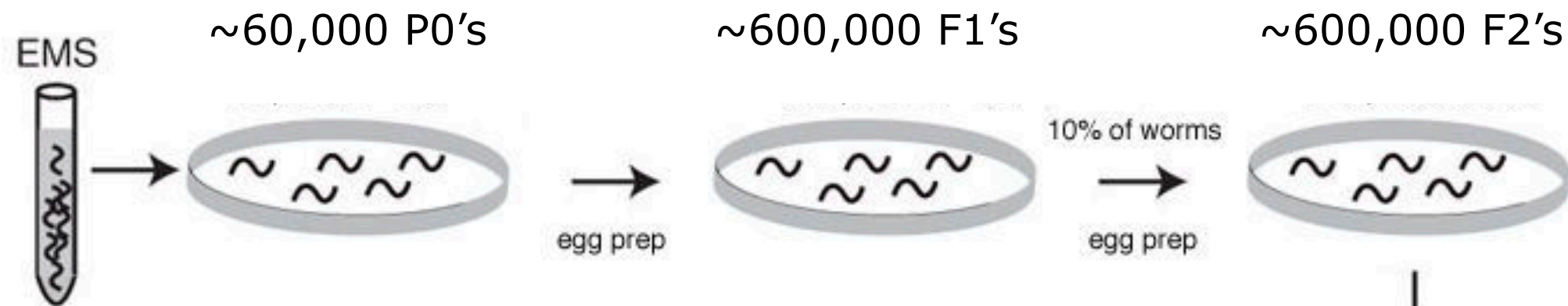
Wormatlas

- Short generation time, 1mm long
- Hermaphrodites (w/ some males)
- Complete wiring diagram of nervous system
- Developmental fate of all 959 somatic cells known, cell position also constant
- Easy to make transgenic animals
- Genome sequenced in >5 closely related species
- Transparency allows for in vivo studies using reporter genes
- 20,470 protein coding genes



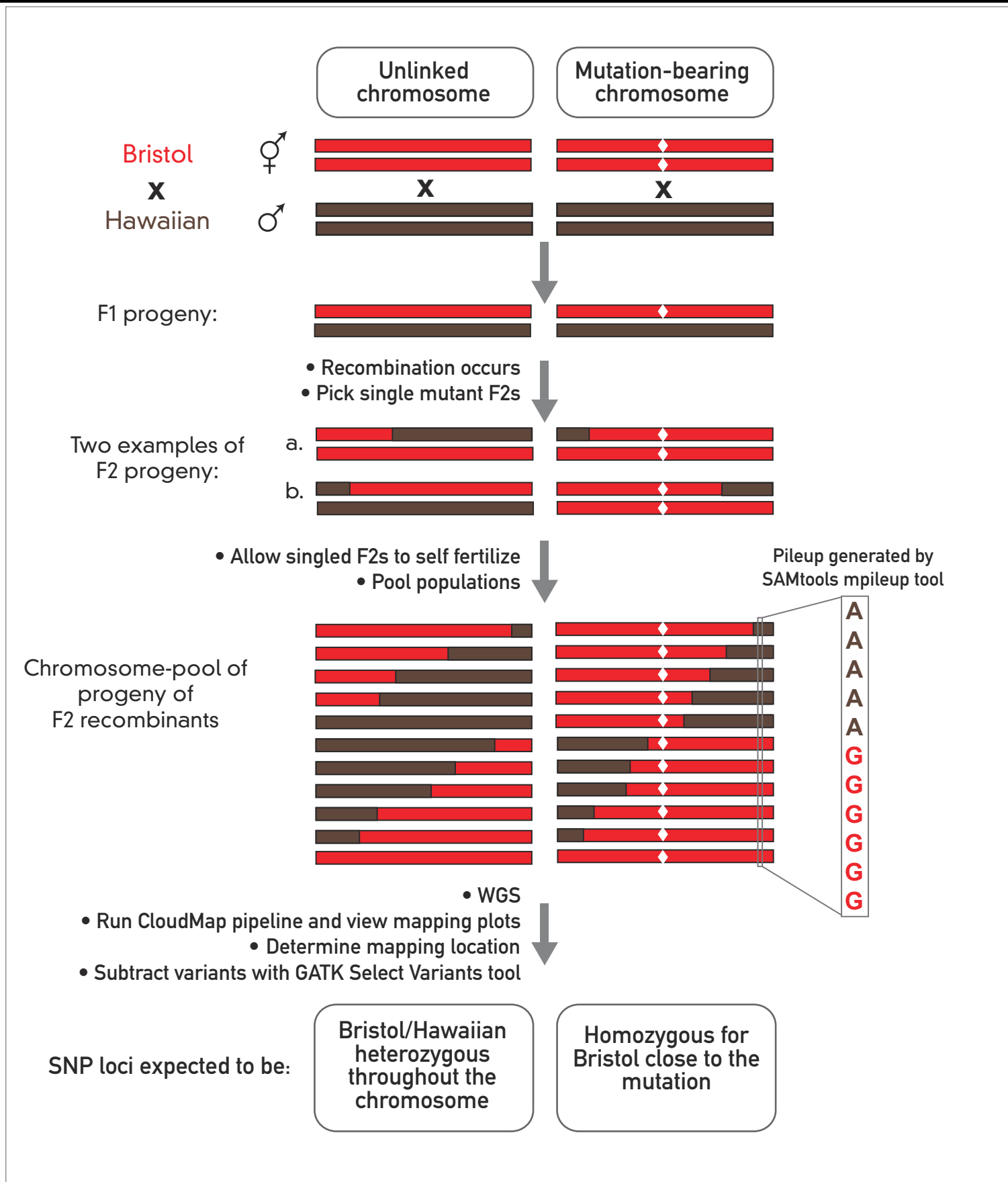
Smith et al. 2010

# Forward genetic screens using a wormsorter



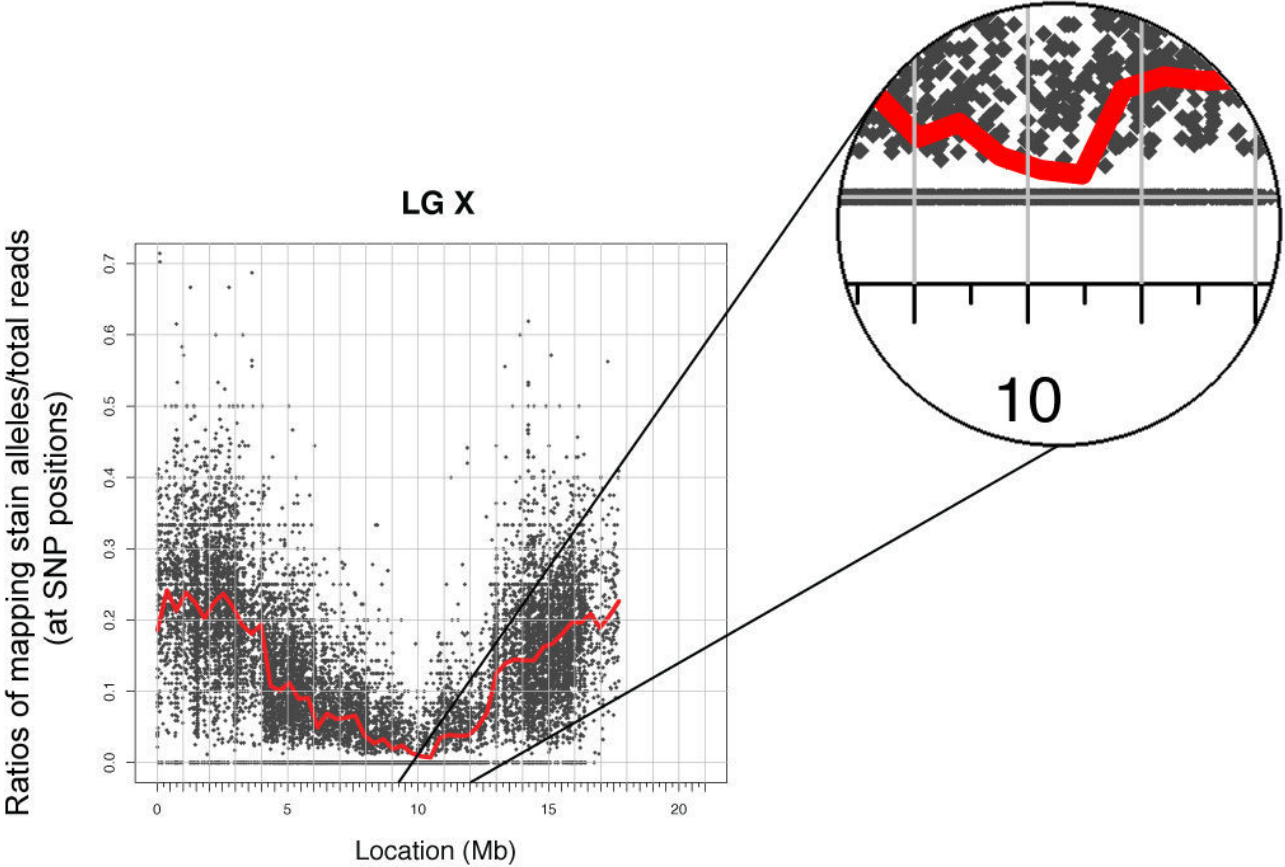
adapted from  
Doitsidou, 2008

# SNP mapping concept

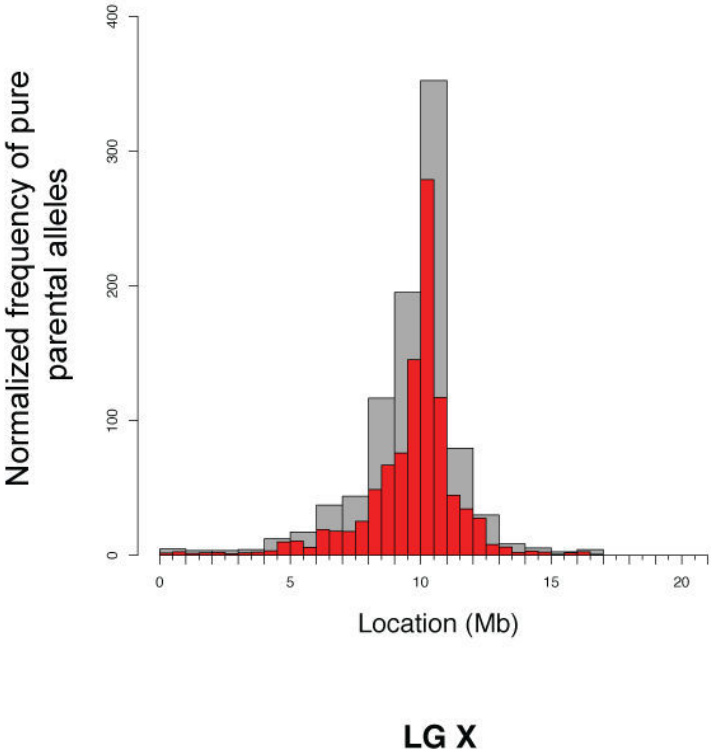




# CloudMap's SNP mapping plots



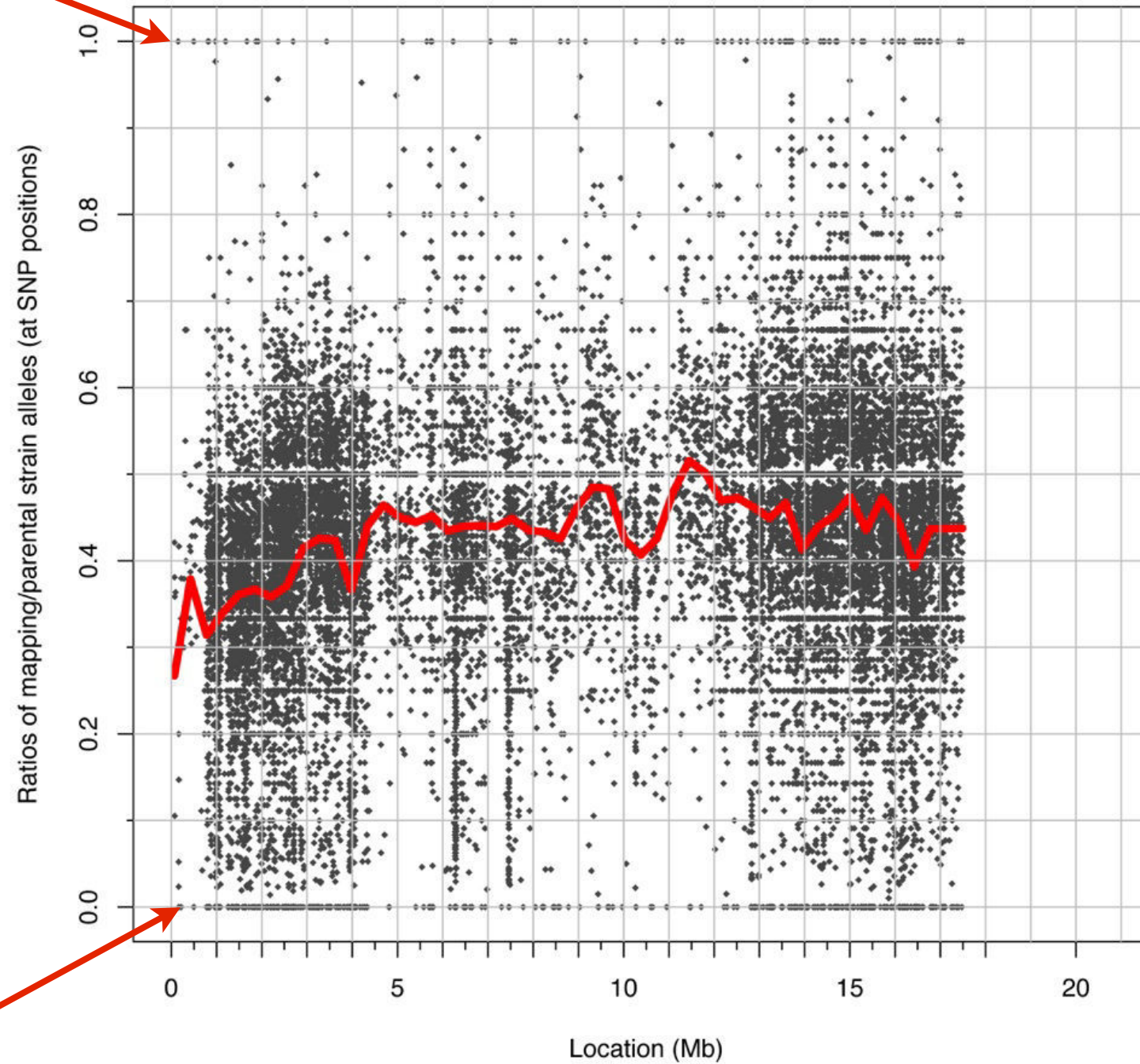
vab-3:LG X 10,503,393-  
10,519,348



# *An unlinked chromosome*

*pure Hawaiian  
ratio*

LG IV

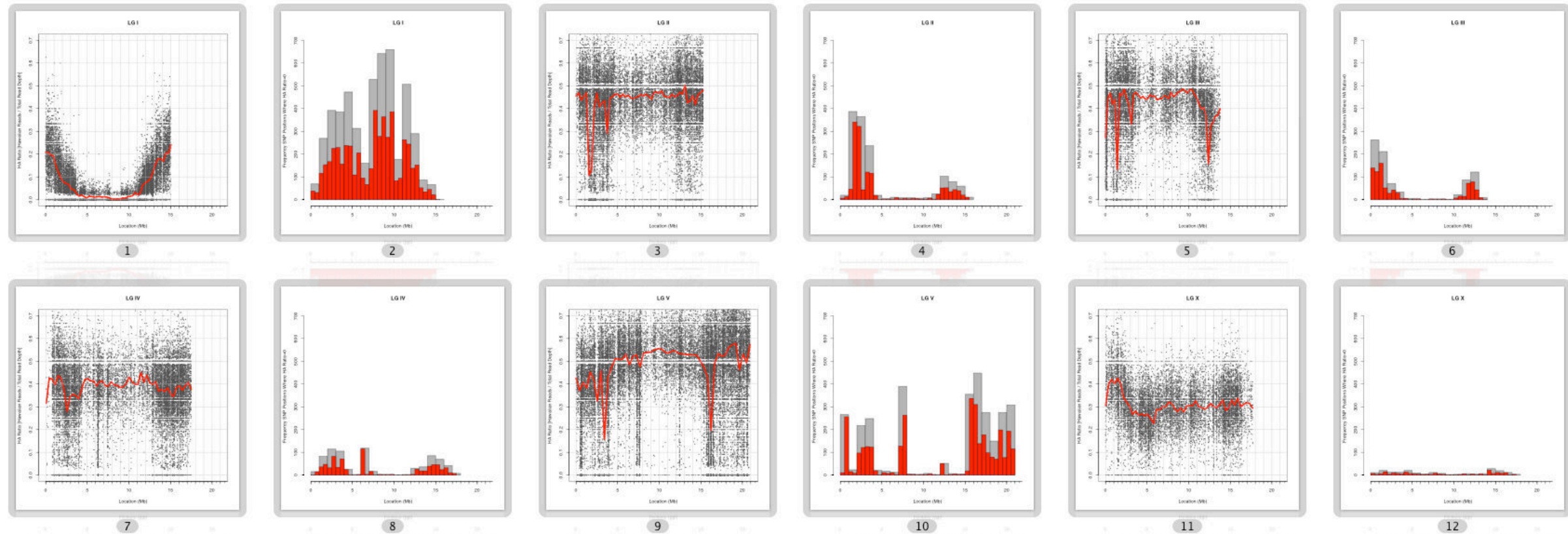


*pure parental  
ratio*

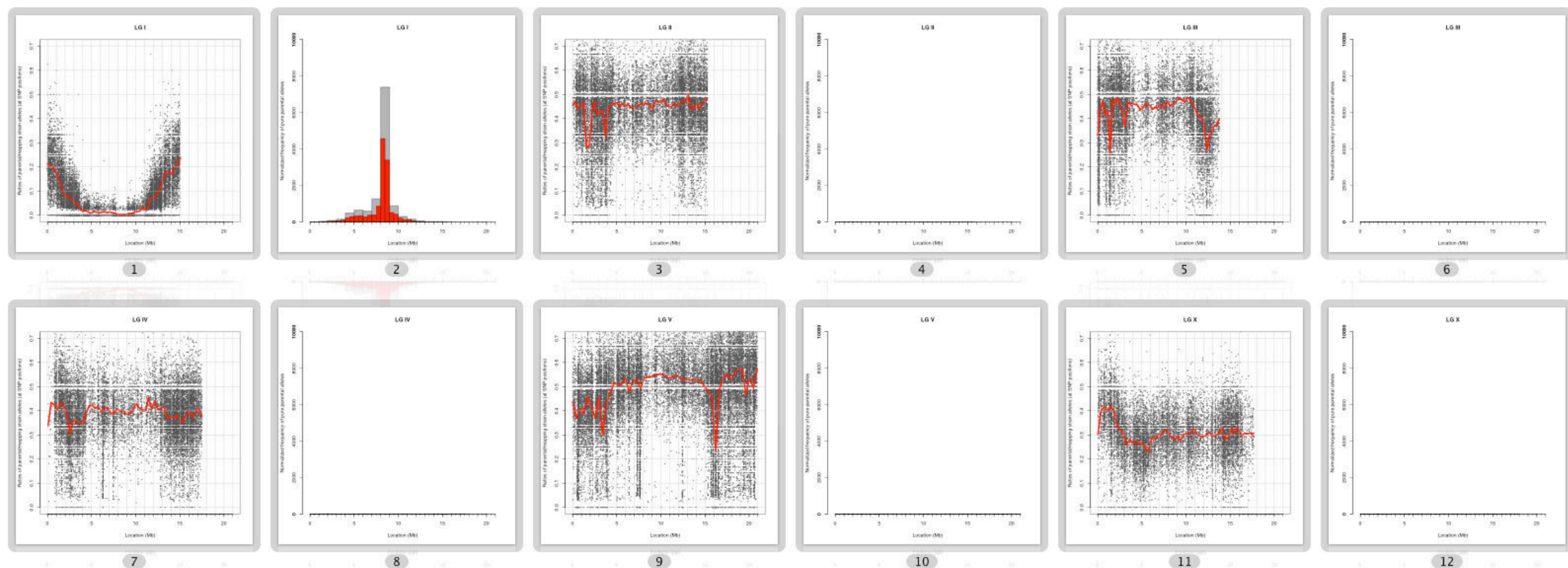


# SNP mapping plots before/after normalization

## Pre-Normalization:

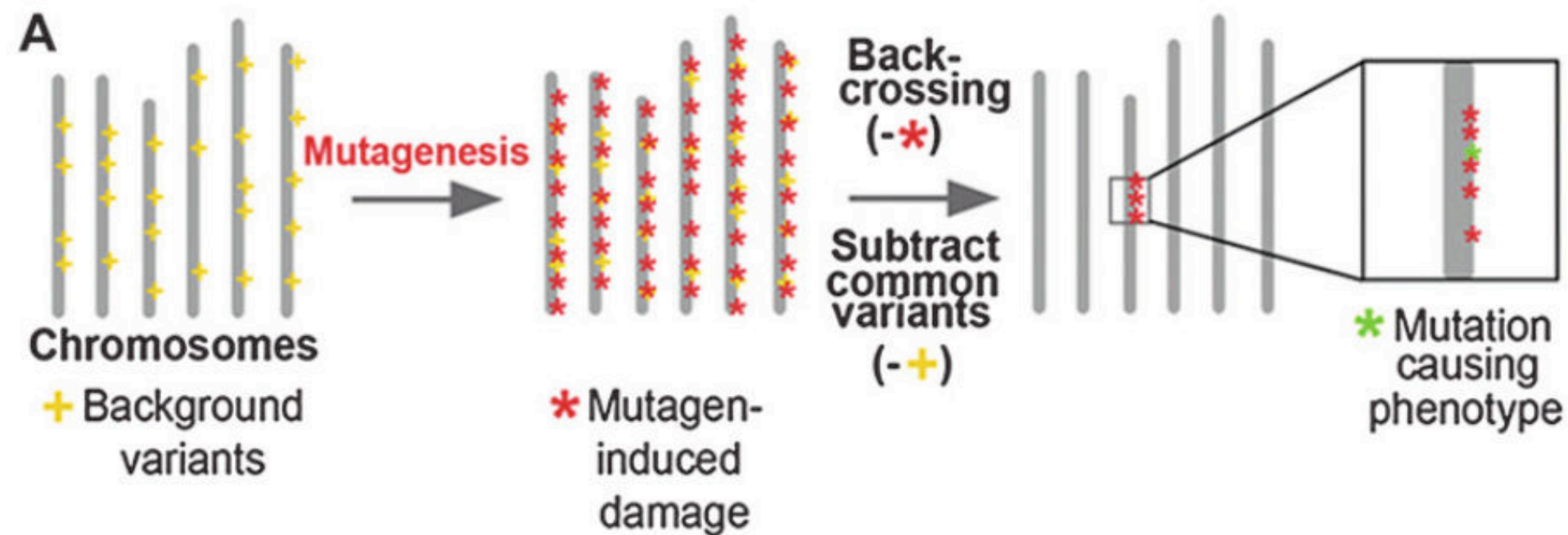


## Post-Normalization:

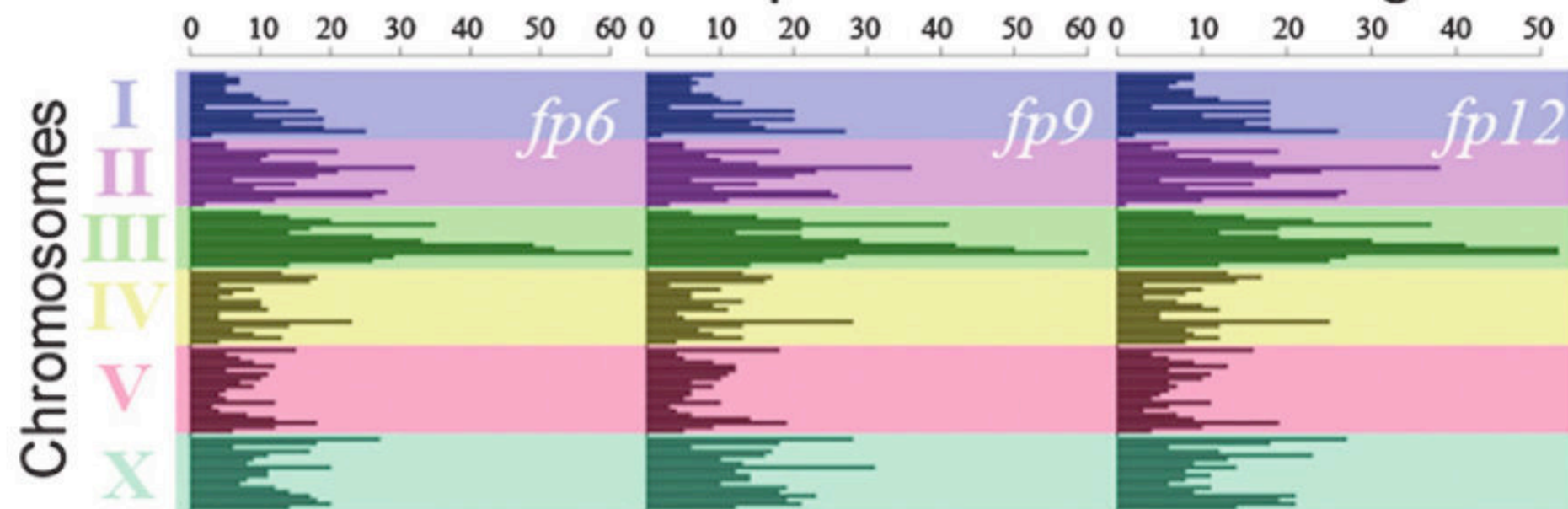




# EMS Density Mapping

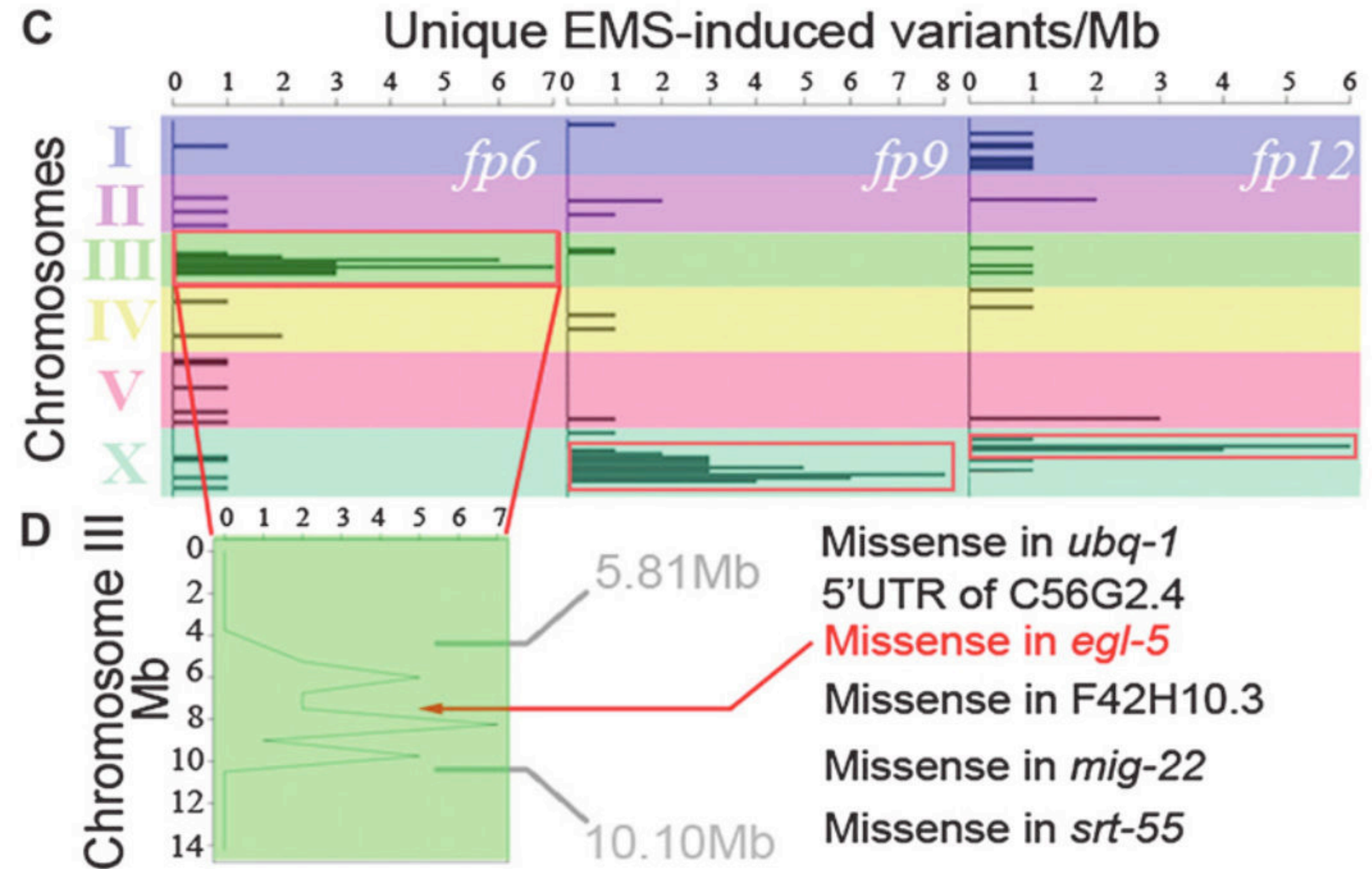


**B** Total variations/Mb compared to N2 reference genome

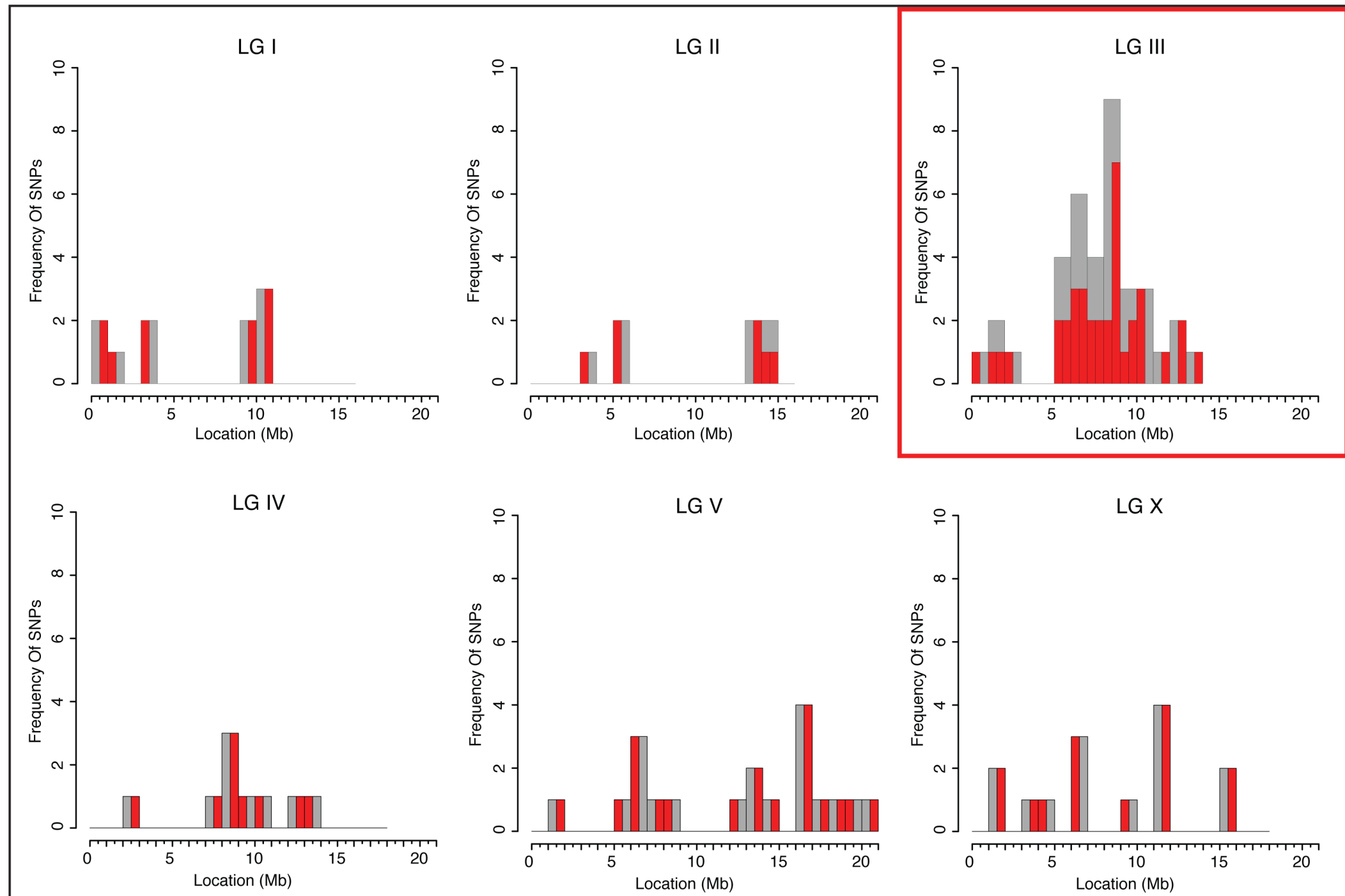


- 1) Subtraction of common variants
- 2) Quality filtering
- 3) Filtering for EMS nucleotide changes

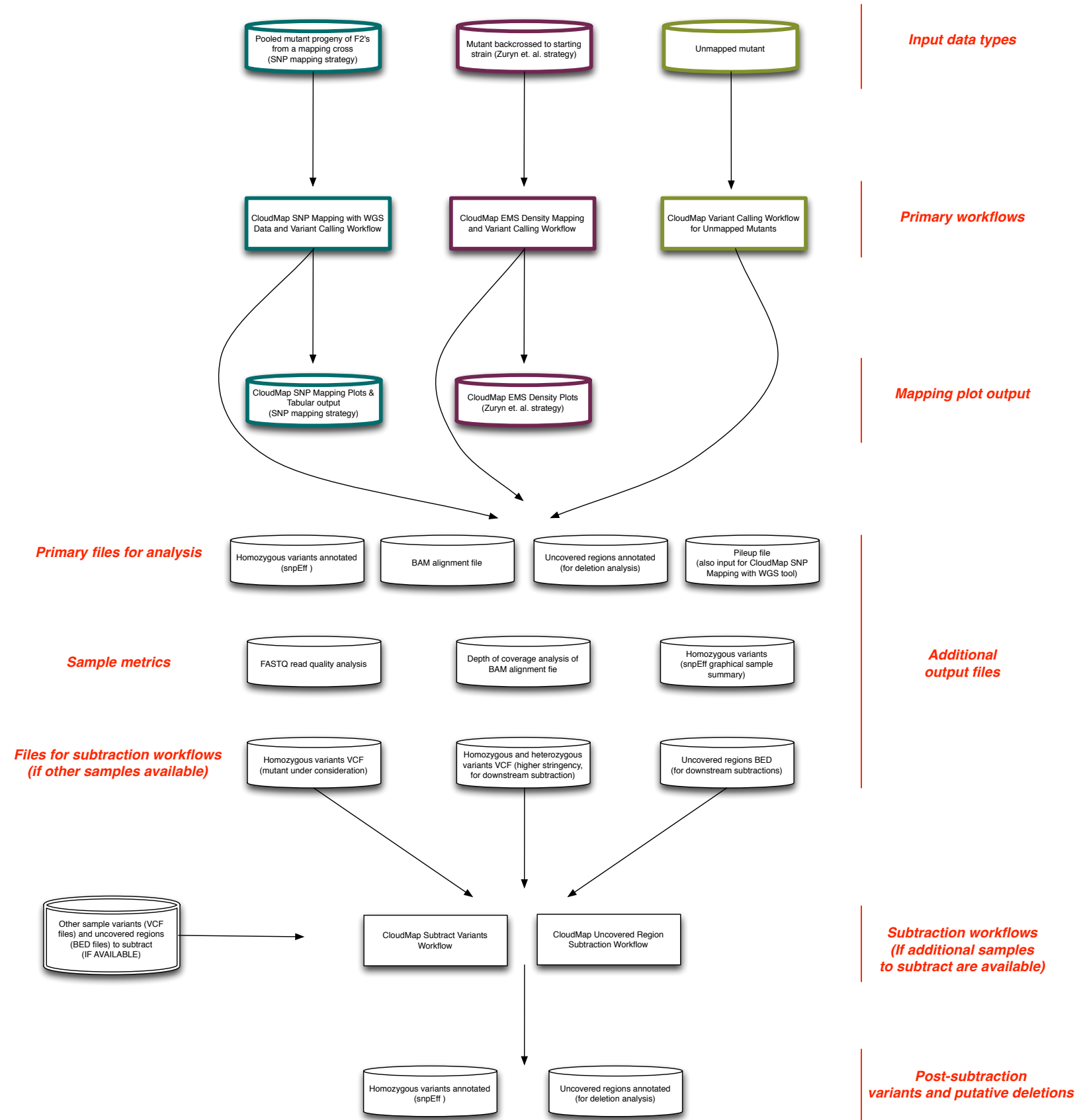
# EMS Density Mapping



# CloudMap: EMS Variants Density Mapping Tool



# CloudMap provides a set of workflows for analysis of mutant genome sequences





*CloudMap provides a set of workflows for analysis of mutant genome sequences*

Galaxy | Published Workflows

https://test.g2.bx.psu.edu/workflow/list\_published

News ▾ C. elegans ▾

**Galaxy** Analyze Data Workflow Shared Data ▾ Visualizat

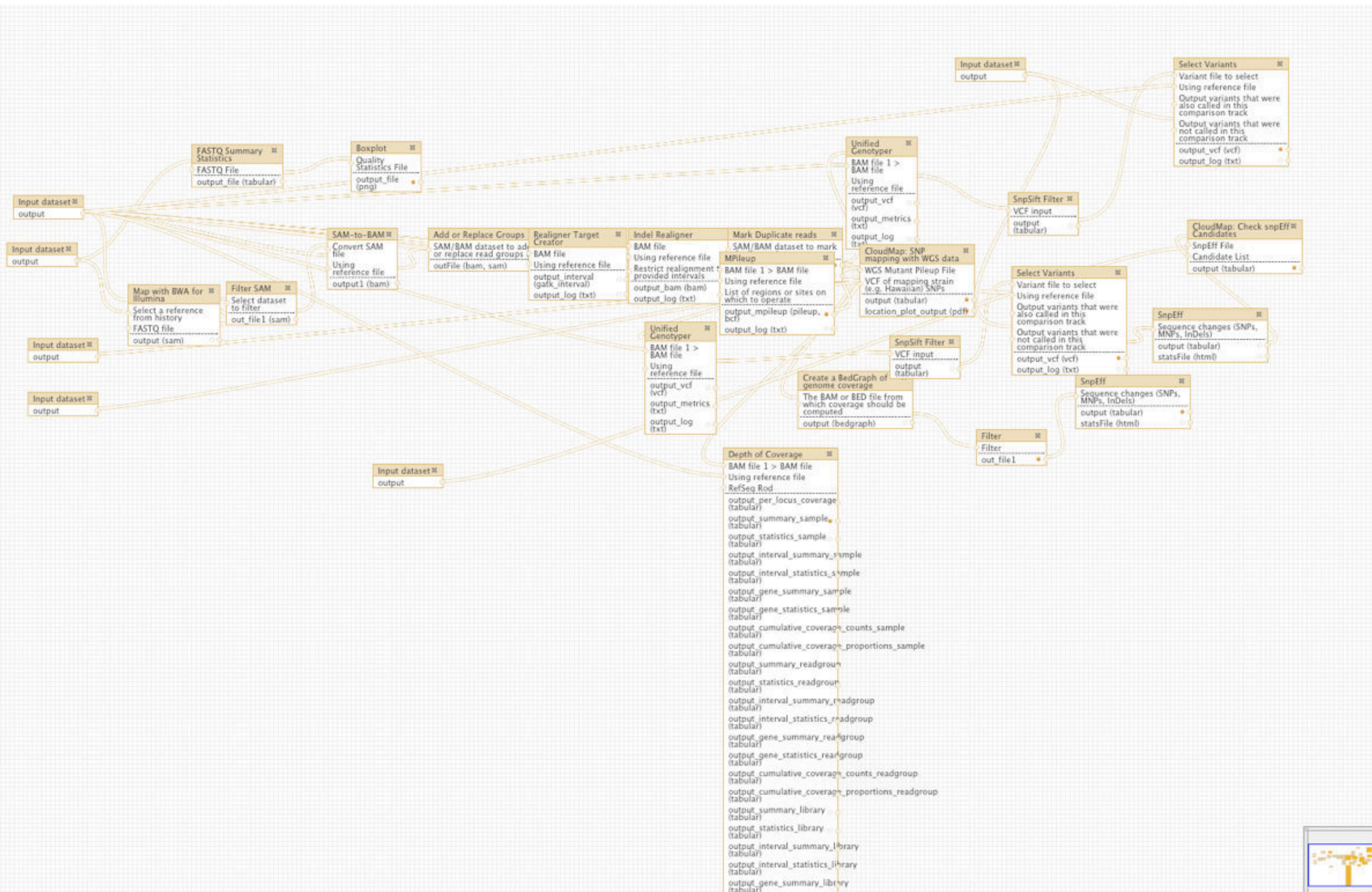
## Published Workflows

cloudmap ✕

[Advanced Search](#)


<u>Name</u>	Annotation	<u>Owner</u>
<a href="#">CloudMap Unmapped Mutant workflow</a>		gal40
<a href="#">CloudMap SNP Mapping with WGS Data and Variant Calling workflow</a>		gal40
<a href="#">Cloudmap Uncovered Region Subtraction workflow</a>		gal40
<a href="#">CloudMap Subtract Variants workflow</a>		gal40

# CloudMap provides a set of workflows for analysis of mutant genome sequences












# CloudMap data libraries contain a proof of principle dataset and config files

 Galaxy

Analyze Data   Workflow   Shared Data ▾   Visualization ▾   Cloud ▾   Help ▾   User ▾

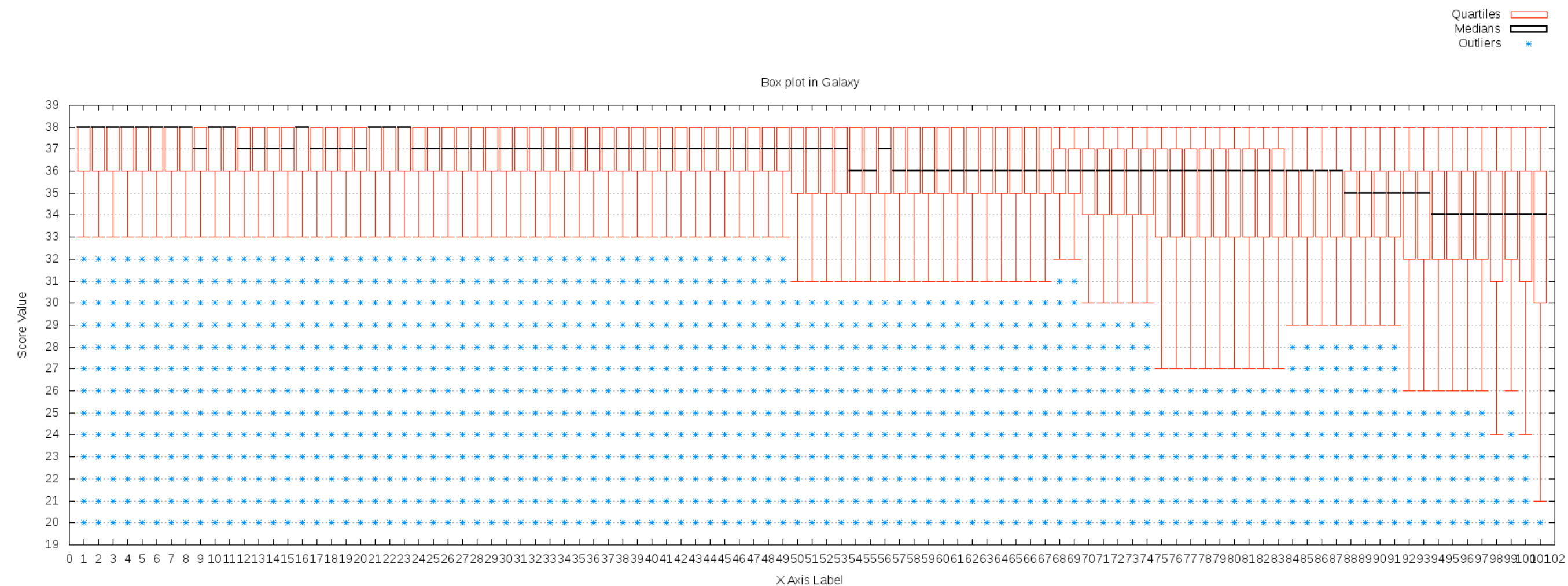
## Data Library "CloudMap"

Contains reference and configuration files for the Cloudmap pipeline

<input type="checkbox"/> Name	Message
<input type="checkbox"/>  Candidate gene lists ▾	Check snpEff output against these candidate genes using CloudMap Check snpEff Candidates tool
<input type="checkbox"/> <input type="checkbox"/> CloudMap_ChromatinFactors.txt ▾	
<input checked="" type="checkbox"/> <input type="checkbox"/> CloudMap_TranscriptionFactors_wTF2.2.txt ▾	List of 937 C. elegans transcription factors from Reece-Hoyes et. al. Nature Methods 2011.
<input type="checkbox"/>  CloudMap user guides ▾	Detailed guides for using the CloudMap pipeline
<input type="checkbox"/>  EMS Variant Density Mapping ▾	Use this dataset to try out the CloudMap EMS Variant Density Mapping tool
<input type="checkbox"/>  ot266 proof of principle dataset ▾	Use these files to run the CloudMap ot266 proof of principle example
<input type="checkbox"/>  Hawaiian SNP reference files unfiltered (WS220.64) ▾	
<input type="checkbox"/> <input type="checkbox"/> HA_SNPS_WS220.64_chr.bed ▾	
<input checked="" type="checkbox"/> <input type="checkbox"/> HA_SNPS_WS220.64_chr.vcf ▾	
<input type="checkbox"/>  ot260 and ot263 VCFs for variant subtraction ▾	Use these VCFs for the CloudMap ot266 proof of principle example
<input checked="" type="checkbox"/> <input type="checkbox"/> ot266_ProofOfPrinciple_Small.fastqsanger ▾	Sample FASTQ file for ot266 Proof of principle
<input checked="" type="checkbox"/> <input type="checkbox"/> HA_SNPs_WS220_Filtered_103626_SNPs_chr.bed ▾	Filtered set of Hawaiian SNP positions (used by mpileup tool)
<input checked="" type="checkbox"/> <input type="checkbox"/> HA_SNPs_WS220_Filtered_103626_SNPs_chr.vcf ▾	Filtered set of Hawaiian SNP variants (used by CloudMap SNP Mapping with WGS tool)
<input checked="" type="checkbox"/> <input type="checkbox"/> WS220.64_chr.fa ▾	WS220.64 genomic reference file
<input type="checkbox"/>  SNP Mapping with WGS Data Other Species Config Files ▾	Use these config files if you want to use the SNP Mapping with WGS Data for any species other than C.elegans and Arabadopsis

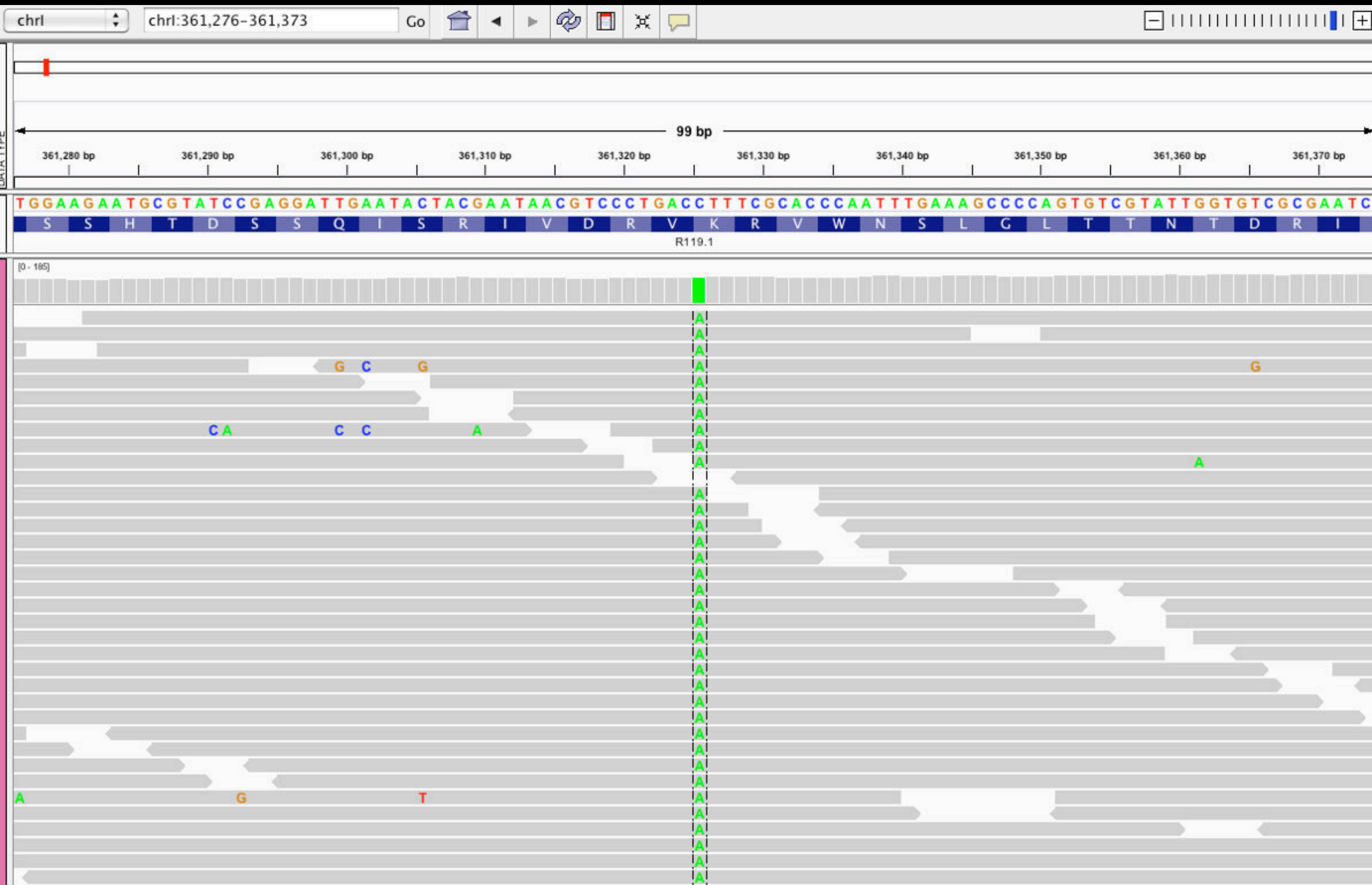
For selected datasets:

# *FASTQ statistics (FASTQ Summary Statistics tool)*





## *Alignments (BWA, GATK realigner, PICARD remove dups)*



# Variant calling (GATK or SAMtools) & variant filtering (snpSift)

	A	B	C	D	E	F	G	H	I	J
1	#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	rgSM
2	chr1	962	.	G	T	367.18	.	AC=1;AF=0.50;AN=2;BaseQRankSum=0.403;DP=23	GT:AD:DP:GQ:PL	0/1:10,13:23:99:397,0,325
3	chr1	991	.	GA	G	100.41	.	AC=1;AF=0.50;AN=2;BaseQRankSum=2.130;DP=14	GT:AD:DP:GQ:PL	0/1:8,6:14:99:139,0,246
4	chr1	1216	.	A	T	68.96	.	AC=1;AF=0.50;AN=2;BaseQRankSum=1.300;DP=7;	GT:AD:DP:GQ:PL	0/1:4,3:7:98.95:99,0,138
5	chr1	1222	.	A	C	109.76	.	AC=1;AF=0.50;AN=2;BaseQRankSum=1.754;DP=7;	GT:AD:DP:GQ:PL	0/1:3,4:7:57.20:140,0,57
6	chr1	1290	.	T	A	126.47	.	AC=1;AF=0.50;AN=2;BaseQRankSum=0.933;DP=14	GT:AD:DP:GQ:PL	0/1:9,5:14:99:156,0,306
7	chr1	1412	.	T	C	235.12	.	AC=1;AF=0.50;AN=2;BaseQRankSum=-1.203;DP=1	GT:AD:DP:GQ:PL	0/1:8,9:17:99:265,0,266
8	chr1	1414	.	G	A	205.1	.	AC=1;AF=0.50;AN=2;BaseQRankSum=-0.209;DP=1	GT:AD:DP:GQ:PL	0/1:7,8:15:99:235,0,233
9	chr1	1421	.	G	A	196.85	.	AC=1;AF=0.50;AN=2;BaseQRankSum=-1.096;DP=1	GT:AD:DP:GQ:PL	0/1:7,8:15:99:227,0,228

# Variant annotation (snpEff)

A	B	C	D	E	F	G	H	J	K	L	M	N	O	P	Q	R	S	T	U
# Chromo	Position	Reference	Change	Change_t	Homozyg	Quality	Coverage	Gene_ID	Gene_name	Bio_type	Transcript	Exon_ID	Exon_Ran	Effect	old_AA/n	Old_codo	Codon_N	Codon_D	CDS_size
I	9879698	C	T	SNP	Het	27	62	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.1			INTRON					1122
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122
I	9909039	G	A	SNP	Het	143	95	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620
I	9909895	G	T	SNP	Het	162	68	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620
I	9910274	*	#NAME?	INS	Het	4.42	102	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620
I	9910270	*	#NAME?	INS	Het	4.42	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620
I	9910274	*	#NAME?	INS	Het	217	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620
I	9994773	A	G	SNP	Het	142	73	T23D8.8	cfi-1	protein_codi	T23D8.8	exon_I_9994	7	NON_SYNON	I/T	aTc/aCc	451	0	1404
I	10139575	C	G	SNP	Het	16.1	28							INTERGENIC					
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkh-10	protein_codi	C25A1.2.2	exon_I_1016	3	SYNONYMOU	H/H	caT/caC	112	1	585
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkh-10	protein_codi	C25A1.2.1	exon_I_1016	3	SYNONYMOU	H/H	caT/caC	112	1	585
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11b			INTRON					1356
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11a			INTRON					1362
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11b	exon_I_1019	5	SYNONYMOU	R/R	cgT/cgA	212	3	1356
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11a	exon_I_1019	5	SYNONYMOU	R/R	cgT/cgA	212	3	1362
I	10207646	A	C	SNP	Het	49	480							INTERGENIC					
I	10209783	A	G	SNP	Hom	27	45							INTERGENIC					
I	10209806	G	A	SNP	Het	3.55	16							INTERGENIC					
I	10248569	G	T	SNP	Het	141	100	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218
I	10248578	C	T	SNP	Het	39	92	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218
I	10251364	T	C	SNP	Het	113	115	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218
I	10251848	C	T	SNP	Het	135	102	ZC247.3	lin-11	protein_codi	ZC247.3	exon_I_1025	5	SYNONYMOU	S/S	tcC/tcT	202	3	1218
I	10383040	G	A	SNP	Het	40	99	F45H11.4	mgl-2	protein_codi	F45H11.4.2			UTR_3_PRIME: 590 bases from CDS					
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3b			INTRON					2406
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3a			INTRON					2454
I	10477650	C	T	SNP	Het	120	83	F37D6.2	F37D6.2	protein_codi	F37D6.2a.1			INTRON					1749

List of variants + Annotation information -> List of variant effects



# CloudMap variant annotation candidate checker

A	B	C	D	E	F	G	H	J	K	L	M	N	O	P	Q	R	S	T	U	Y	Z
# Chromo	Position	Reference	Change	Change_t	Homozyg	Quality	Coverage	Gene_ID	Gene_nar	Bio_type	Transcript	Exon_ID	Exon_Ran	Effect	old_AA/n	Old_codo	Codon_N	Codon_D	CDS_size	TFs	
I	9879698	C	T	SNP	Het	27	62	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.1			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9909039	G	A	SNP	Het	143	95	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9909895	G	T	SNP	Het	162	68	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	4.42	102	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910270	*	#NAME?	INS	Het	4.42	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	217	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9994773	A	G	SNP	Het	142	73	T23D8.8	cfi-1	protein_codi	T23D8.8	exon_I_9994		7 NON_SYNON I/T		aTc/aCc		451	0	1404	ARID/BRIGHT
I	10139575	C	G	SNP	Het	16.1	28							INTERGENIC							WH - Fork Head, AT Hook
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.2	exon_I_1016		3 SYNONYMOU H/H		caT/caC		112	1	585	WH - Fork Head
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.1	exon_I_1016		3 SYNONYMOU H/H		caT/caC		112	1	585	WH - Fork Head
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11b			INTRON						1356	bHLH
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11a			INTRON						1362	bHLH
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11b	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA		212	3	1356	bHLH
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11a	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA		212	3	1362	bHLH
I	10207646	A	C	SNP	Het	49	480							INTERGENIC							WH - Fork Head, AT Hook
I	10209783	A	G	SNP	Hom	27	45							INTERGENIC							WH - Fork Head, AT Hook
I	10209806	G	A	SNP	Het	3.55	16							INTERGENIC							WH - Fork Head, AT Hook
I	10248569	G	T	SNP	Het	141	100	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON						1218	HD - LIM
I	10248578	C	T	SNP	Het	39	92	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON						1218	HD - LIM
I	10251364	T	C	SNP	Het	113	115	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON						1218	HD - LIM
I	10251848	C	T	SNP	Het	135	102	ZC247.3	lin-11	protein_codi	ZC247.3	exon_I_1025		5 SYNONYMOU S/S		tcC/tcT		202	3	1218	HD - LIM
I	10383040	G	A	SNP	Het	40	99	F45H11.4	mgl-2	protein_codi	F45H11.4.2			UTR_3_PRIME: 590 bases from CDS							bZIP
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3b			INTRON						2406	ZF - C2H2 - 4 fingers
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3a			INTRON						2454	ZF - C2H2 - 4 fingers
I	10477650	C	T	SNP	Het	120	83	F37D6.2	F37D6.2	protein_codi	F37D6.2a.1			INTRON						1749	ZF - C2H2 - 5 fingers

1) Transcription factors

2) Transgene silencers

3) Genes expressed in the nervous system

4) Anything you want. . .



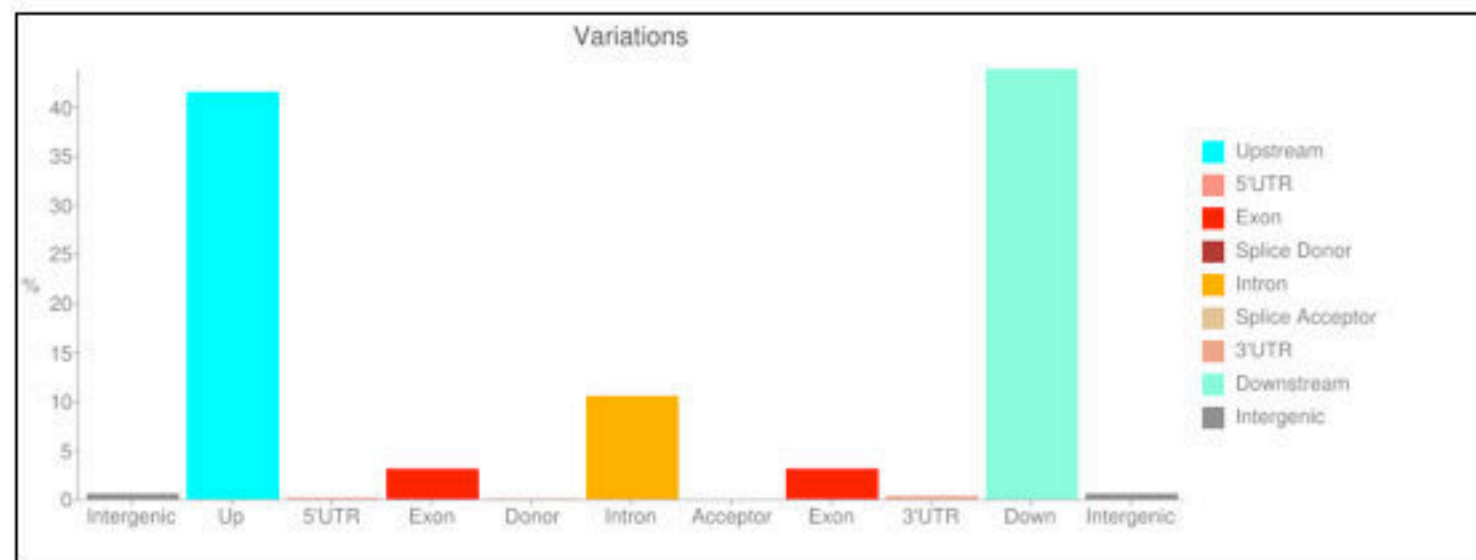
# Variant annotation (snpEff)

Number of effects by impact

Type (alphabetical order)	Count	Percent
HIGH	245	0.774%
LOW	16,895	53.371%
MODERATE	501	1.583%
MODIFIER	14,015	44.273%

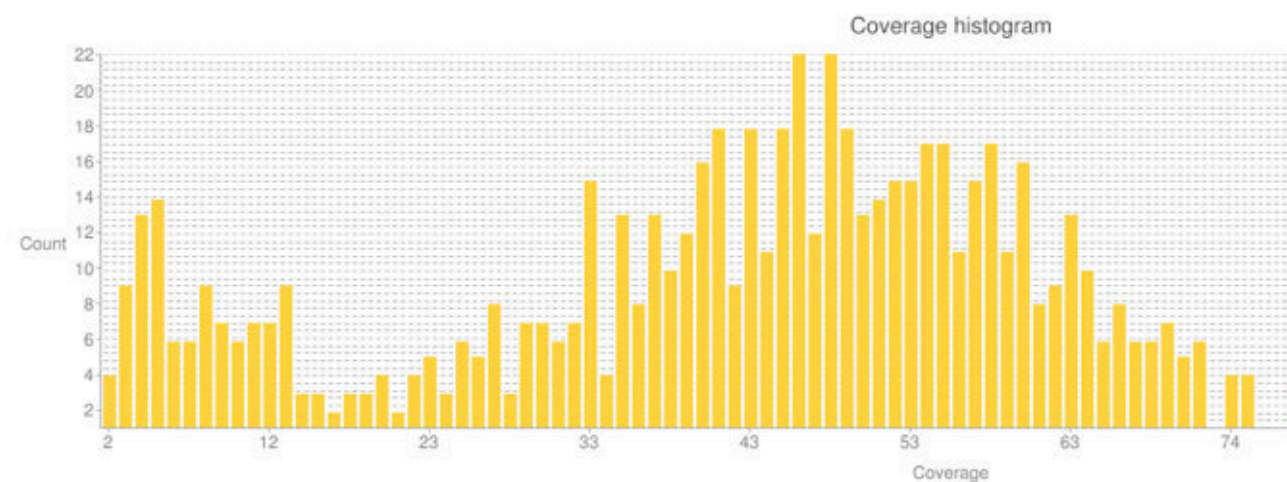
Number of effects by type and region

Type			Region		
Type (alphabetical order)	Count	Percent	Type (alphabetical order)	Count	Percent
CODON_CHANGE_PLUS_CODON_DELETION	1	0.003%	DOWNSTREAM	13,868	43.808%
CODON_CHANGE_PLUS_CODON_INSERTION	1	0.003%	EXON	983	3.105%
CODON_DELETION	2	0.006%	INTERGENIC	177	0.559%
CODON_INSERTION	4	0.013%	INTRON	3,332	10.526%
DOWNSTREAM	13,868	43.808%	SPLICE_SITE_ACCEPTOR	6	0.019%
FRAME_SHIFT	211	0.667%	SPLICE_SITE_DONOR	9	0.028%
INTERGENIC	177	0.559%	UPSTREAM	13,134	41.49%
INTRON	3,332	10.526%	UTR_3_PRIME	104	0.329%
NON_SYNONYMOUS_CODING	493	1.557%	UTR_5_PRIME	43	0.136%
SPLICE_SITE_ACCEPTOR	6	0.019%			
SPLICE_SITE_DONOR	9	0.028%			
STOP_GAINED	15	0.047%			
STOP_LOST	4	0.013%			
SYNONYMOUS_CODING	250	0.79%			
SYNONYMOUS_START	1	0.003%			
SYNONYMOUS_STOP	1	0.003%			
UPSTREAM	13,134	41.49%			
UTR_3_PRIME	104	0.329%			
UTR_5_PRIME	43	0.136%			

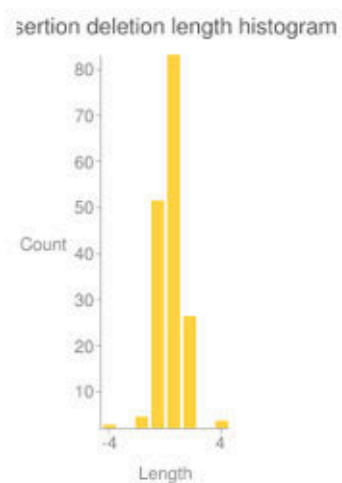


## Variant annotation (snpEff)

Coverage:																																																					
Min	2																																																				
Max	1,370																																																				
Mean	54.308																																																				
Median	46																																																				
Standard deviation	91.976																																																				
Values	2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 20 21 22 23 24 25 26 27 28 29 30 31 32 33 34 35 36 37 38 39 40 41 42 43 44 45 46 47 48 49 50 51 52																																																				
Count	4 9 13 14 6 6 9 7 6 7 7 9 3 3 2 3 3 4 2 4 5 3 6 5 8 3 7 7 6 7 15 4 13 8 13 10 12 16 18 9 18 11 18 22 12 22 18 13 14 15 15 17 17 11 15 17 11 16 8 9																																																				



Insertions and deletions length:	
Min	-4
Max	4
Mean	0.444
Median	1
Standard deviation	1.457
Values	-4 -3 -2 -1 1 2 3 4
Count	3 2 5 52 83 27 2 4



# Variant annotation (snpEff)

## Base changes (SNPs)

	A	C	G	T
A	0	17	17	28
C	13	0	11	183
G	172	14	0	20
T	29	19	10	0

## Ts/Tv (transitions / transversions)

**Note:** Only SNPs are used for this statistic.

**Note:** This Ts/Tv ratio is a 'raw' ratio. Some people prefer to use a ratio of rates, not observed events. In that case, you need to multiply by 2.0 (since there are twice as many possible transitions than transversions, E[Ts/Tv] ratio is twice the ratio of events).

Transitions	391
Transversions	142
Ts/Tv ratio	2.7535

## All variants:

Sample	:	Total
Transitions	:	391 391
Transversions	:	142 142
Ts/Tv	:	2.754 2.754

**Only known variants** (i.e. the ones having a non-empty ID field):

No results available (empty input?)

## Variant annotation (snpEff)

### Amino acid changes

How to read this table:

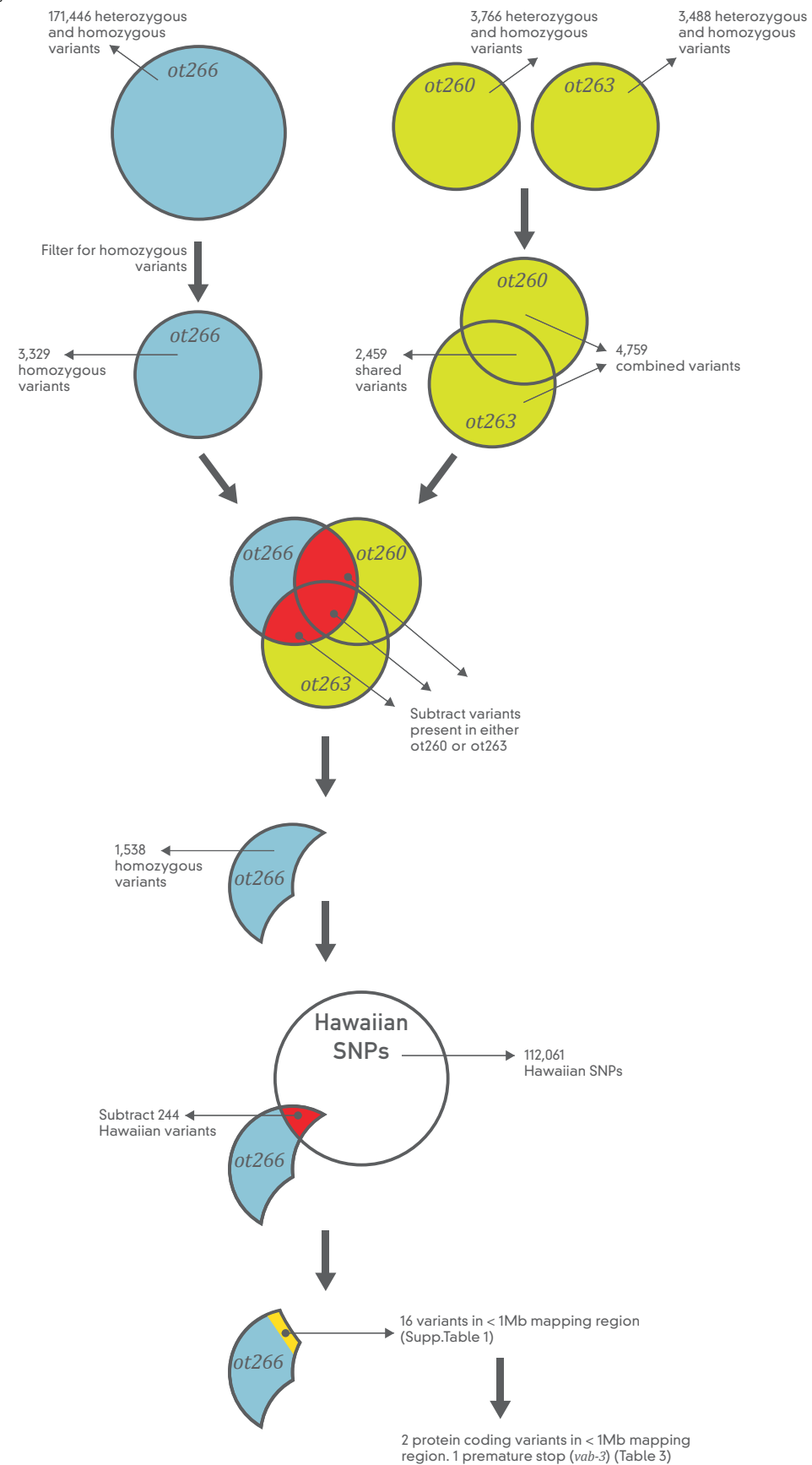
- Rows are reference amino acids and columns are changed amino acids. E.g. Row 'A' column 'E' indicates how many 'A' amino acids have been replaced by 'E' amino acids.
- Red background colors indicate that more changes happened (heat-map).
- Diagonals are indicated using grey background color
- WARNING: This table may include different translation codon tables (e.g. mamalian DNA and mitochondrial DNA).

[illegible]



# Variant subtraction and filtration (GATK)

Fig.8



# In silico complementation testing (compare two datasets tool)

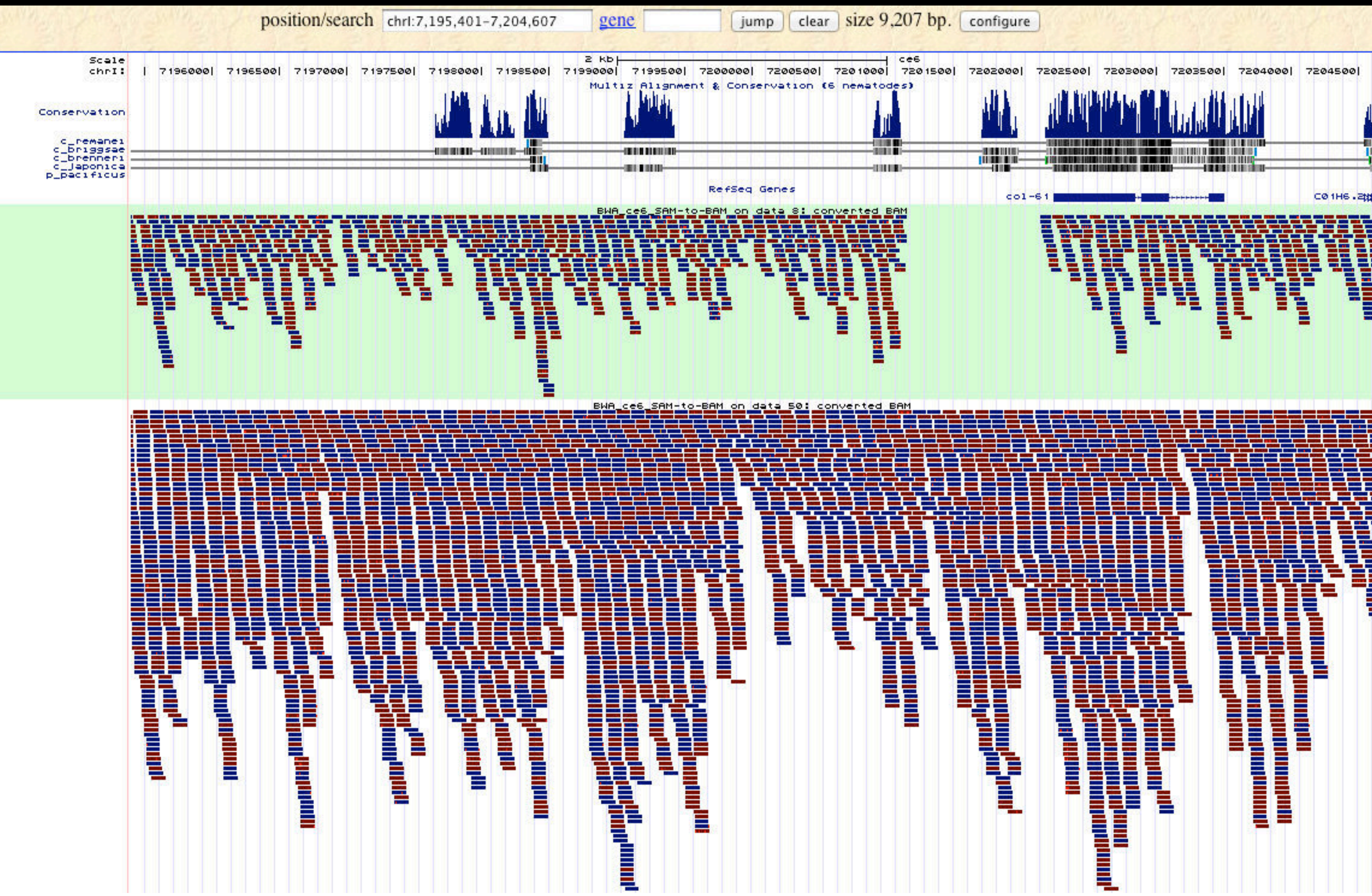
A	B	C	D	E	F	G	H	J	K	L	M	N	O	P	Q	R	S	T	U	Y	Z
# Chromo	Position	Reference	Change	Change_t	Homozyg	Quality	Coverage	Gene_ID	Gene_nar	Bio_type	Transcript	Exon_ID	Exon_Ran	Effect	old_AA/n	Old_codo	Codon_N	Codon_D	CDS_size	TFs	
I	9879698	C	T	SNP	Het	27	62	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.1			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9909039	G	A	SNP	Het	143	95	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9909895	G	T	SNP	Het	162	68	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	4.42	102	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910270	*	#NAME?	INS	Het	4.42	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	217	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9994773	A	G	SNP	Het	142	73	T23D8.8	cfi-1	protein_codi	T23D8.8	exon_I_9994		7 NON_SYNON I/T		aTc/aCc	451	0	1404	ARID/BRIGHT	
I	10139575	C	G	SNP	Het	16.1	28							INTERGENIC						WH - Fork Head, AT Hook	
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.2	exon_I_1016		3 SYNONYMOU H/H		caT/caC	112	1	585	WH - Fork Head	
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.1	exon_I_1016		3 SYNONYMOU H/H		caT/caC	112	1	585	WH - Fork Head	
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11b			INTRON					1356	bHLH	
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11a			INTRON					1362	bHLH	
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11b	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA	212	3	1356	bHLH	
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11a	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA	212	3	1362	bHLH	
I	10207646	A	C	SNP	Het	49	480							INTERGENIC						WH - Fork Head, AT Hook	
I	10209783	A	G	SNP	Hom	27	45							INTERGENIC						WH - Fork Head, AT Hook	
I	10209806	G	A	SNP	Het	3.55	16							INTERGENIC						WH - Fork Head, AT Hook	
I	10248569	G	T	SNP	Het	141	100	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10248578	C	T	SNP	Het	39	92	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10251364	T	C	SNP	Het	113	115	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10251848	C	T	SNP	Het	135	102	ZC247.3	lin-11	protein_codi	ZC247.3	exon_I_1025		5 SYNONYMOU S/S		tcC/tcT	202	3	1218	HD - LIM	
I	10383040	G	A	SNP	Het	40	99	F45H11.4	mgl-2	protein_codi	F45H11.4.2			UTR_3_PRIME: 590 bases from CDS						bZIP	
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3b			INTRON					2406	ZF - C2H2 - 4 fingers	
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3a			INTRON					2454	ZF - C2H2 - 4 fingers	
I	10477650	C	T	SNP	Het	120	83	F37D6.2	F37D6.2	protein_codi	F37D6.2a.1			INTRON					1749	ZF - C2H2 - 5 fingers	

What genes have been hit in both members of a complementation group yet have different mutations?

A	B	C	D	E	F	G	H	J	K	L	M	N	O	P	Q	R	S	T	U	Y	Z
# Chromo	Position	Reference	Change	Change_t	Homozyg	Quality	Coverage	Gene_ID	Gene_nar	Bio_type	Transcript	Exon_ID	Exon_Ran	Effect	old_AA/n	Old_codo	Codon_N	Codon_D	CDS_size	TFs	
I	9879698	C	T	SNP	Het	27	62	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.1			INTRON					1122	ZF - NHR	
I	9880489	C	T	SNP	Het	192	109	T23H4.2	nhr-69	protein_codi	T23H4.2.2			INTRON					1122	ZF - NHR	
I	9909039	G	A	SNP	Het	143	95	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9909895	G	T	SNP	Het	162	68	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	4.42	102	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910270	*	#NAME?	INS	Het	4.42	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9910274	*	#NAME?	INS	Het	217	103	F52F12.6	ztf-11	protein_codi	F52F12.6			INTRON					1620	ZF - C2HC 2 fingers	
I	9994773	A	G	SNP	Het	142	73	T23D8.8	cfi-1	protein_codi	T23D8.8	exon_I_9994		7 NON_SYNON I/T		aTc/aCc	451	0	1404	ARID/BRIGHT	
I	10139575	C	G	SNP	Het	16.1	28							INTERGENIC						WH - Fork Head, AT Hook	
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.2	exon_I_1016		3 SYNONYMOU H/H		caT/caC	112	1	585	WH - Fork Head	
I	10163982	A	G	SNP	Het	182	125	C25A1.2	fkx-10	protein_codi	C25A1.2.1	exon_I_1016		3 SYNONYMOU H/H		caT/caC	112	1	585	WH - Fork Head	
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11b			INTRON					1356	bHLH	
I	10194812	G	A	SNP	Het	133	79	C25A1.11	aha-1	protein_codi	C25A1.11a			INTRON					1362	bHLH	
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11b	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA	212	3	1356	bHLH	
I	10195403	A	T	SNP	Het	222	147	C25A1.11	aha-1	protein_codi	C25A1.11a	exon_I_1019		5 SYNONYMOU R/R		cgT/cgA	212	3	1362	bHLH	
I	10207646	A	C	SNP	Het	49	480							INTERGENIC						WH - Fork Head, AT Hook	
I	10209783	A	G	SNP	Hom	27	45							INTERGENIC						WH - Fork Head, AT Hook	
I	10209806	G	A	SNP	Het	3.55	16							INTERGENIC						WH - Fork Head, AT Hook	
I	10248569	G	T	SNP	Het	141	100	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10248578	C	T	SNP	Het	39	92	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10251364	T	C	SNP	Het	113	115	ZC247.3	lin-11	protein_codi	ZC247.3			INTRON					1218	HD - LIM	
I	10251848	C	T	SNP	Het	135	102	ZC247.3	lin-11	protein_codi	ZC247.3	exon_I_1025		5 SYNONYMOU S/S		tcC/tcT	202	3	1218	HD - LIM	
I	10383040	G	A	SNP	Het	40	99	F45H11.4	mgl-2	protein_codi	F45H11.4.2			UTR_3_PRIME: 590 bases from CDS						bZIP	
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3b			INTRON					2406	ZF - C2H2 - 4 fingers	
I	10411503	A	G	SNP	Het	218	77	F25D7.3	blmp-1	protein_codi	F25D7.3a			INTRON					2454	ZF - C2H2 - 4 fingers	
I	10477650	C	T	SNP	Het	120	83	F37D6.2	F37D6.2	protein_codi	F37D6.2a.1			INTRON					1749	ZF - C2H2 - 5 fingers	



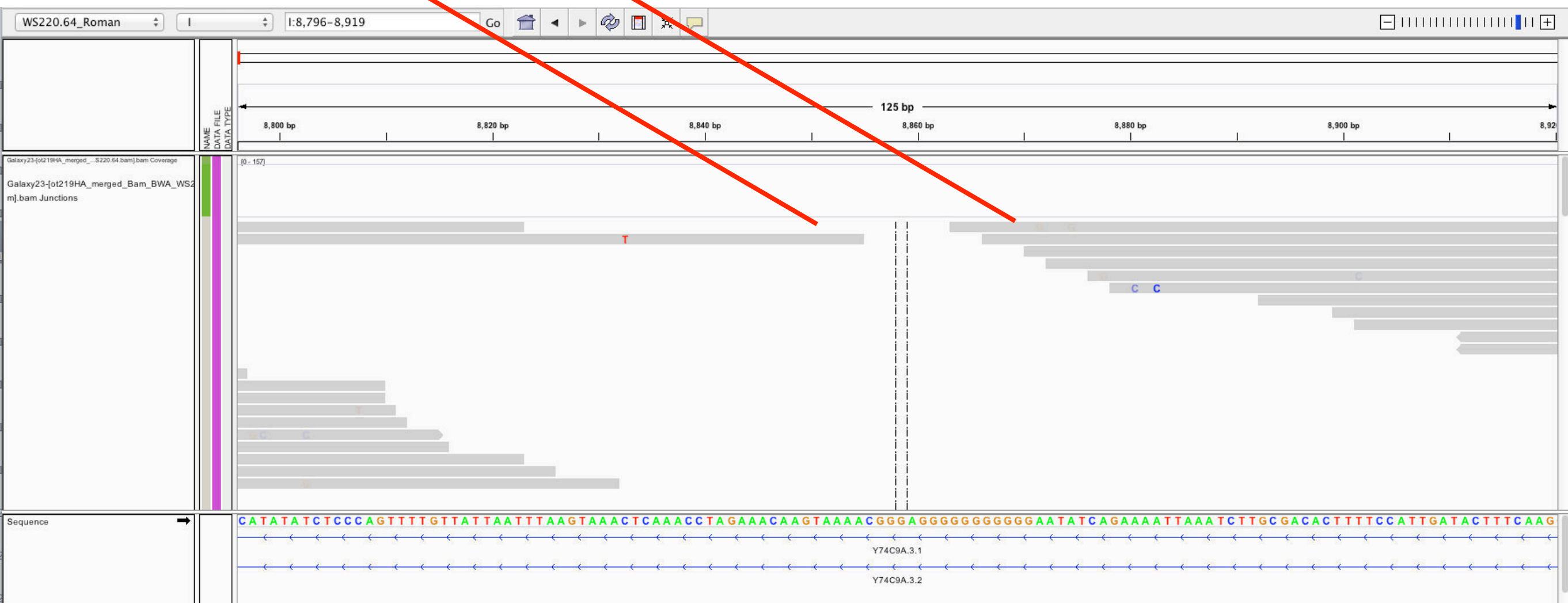
# Which uncovered regions are deletions?





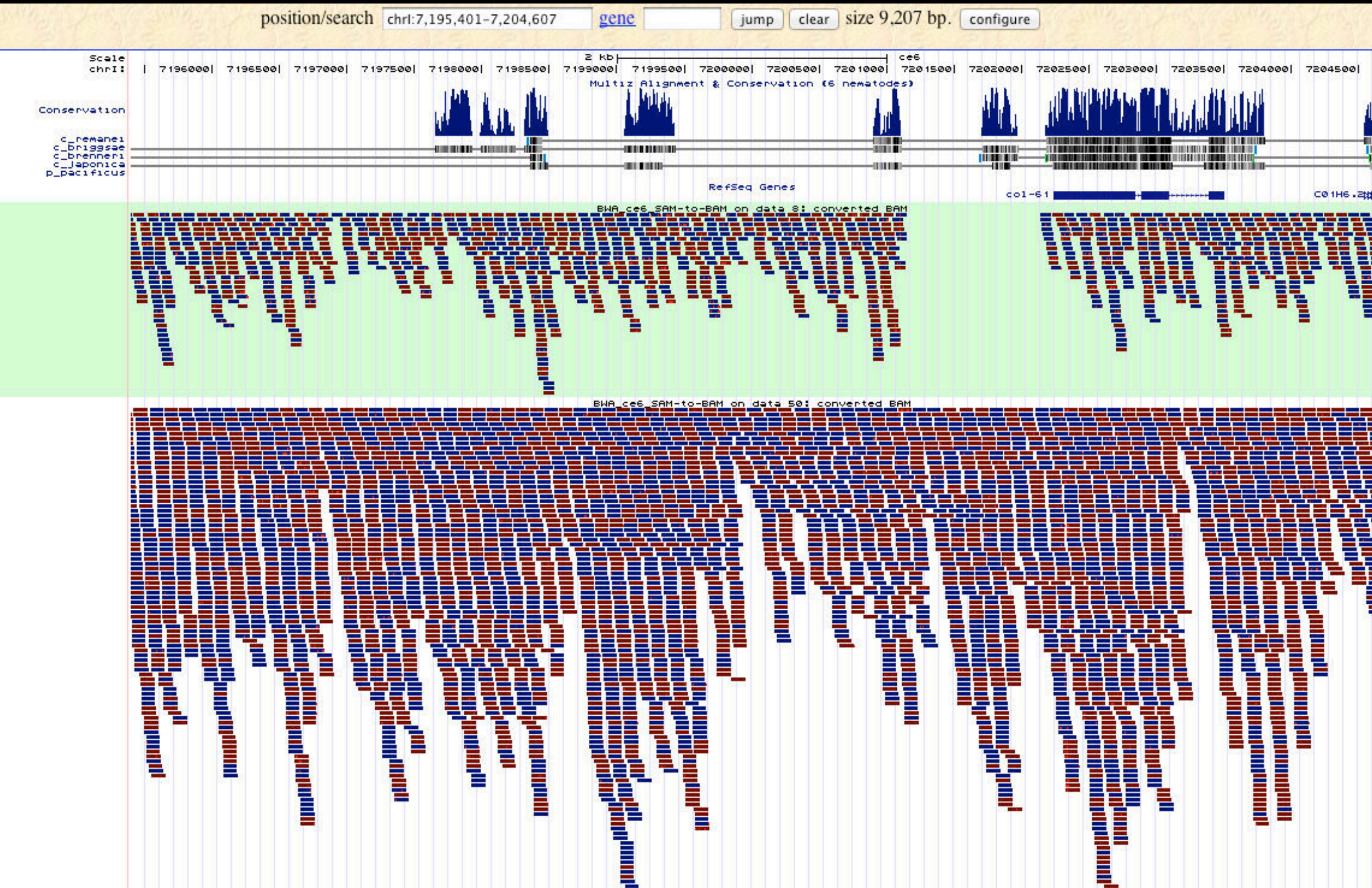
# Identifying unique uncovered regions

	A	B	C	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S	T	U	V	W	X
1	# Chromo	Position	Reference	Homozygous	Coverage	Warnings	Gene_ID	Gene_name	Bio_type	Transcript_ID	Exon_ID	Exon_Rank	Effect	old_AA/new	Old_codon/h	Codon_Num	Codon_Dege	CDS_size	Codons_around	AAs_around	Custom_interval_ID		
2	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.6			UPSTREAM: 2763 bases								0	
3	I	8855	8862	Interval	0	0		Y74C9A.6	Y74C9A.6	snoRNA	Y74C9A.6			UPSTREAM: 4946 bases								0	
4	I	8855	8862	Interval	0	0		Y74C9A.3	Y74C9A.3	protein_codi	Y74C9A.3.1			INTRON					705			0	
5	I	8855	8862	Interval	0	0		Y74C9A.3	Y74C9A.3	protein_codi	Y74C9A.3.2			INTRON					705			0	
6	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.3			UPSTREAM: 1558 bases								0	
7	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.2			UPSTREAM: 2640 bases								0	
8	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.1			UPSTREAM: 2644 bases								0	
9	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.5			UPSTREAM: 2644 bases								0	
10	I	8855	8862	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.4			UPSTREAM: 2650 bases								0	
11	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.6			UPSTREAM: 370 bases								0	
12	I	11248	11266	Interval	0	0		Y74C9A.3	Y74C9A.3	protein_codi	Y74C9A.3.2			UPSTREAM: 1018 bases								0	
13	I	11248	11266	Interval	0	0		Y74C9A.3	Y74C9A.3	protein_codi	Y74C9A.3.1			UPSTREAM: 1016 bases								0	
14	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.3			INTRON					372			0	
15	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.2			UPSTREAM: 247 bases								0	
16	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.1			UPSTREAM: 251 bases								0	
17	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.5			UPSTREAM: 251 bases								0	
18	I	11248	11266	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.4			UPSTREAM: 257 bases								0	
19	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.2			INTRON					372			0	
20	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.1			INTRON					372			0	
21	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.4			INTRON					372			0	
22	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.3			INTRON					372			0	
23	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.6			INTRON					372			0	
24	I	15671	15712	Interval	0	0		Y74C9A.2	nlp-40	protein_codi	Y74C9A.2.5			INTRON					372			0	





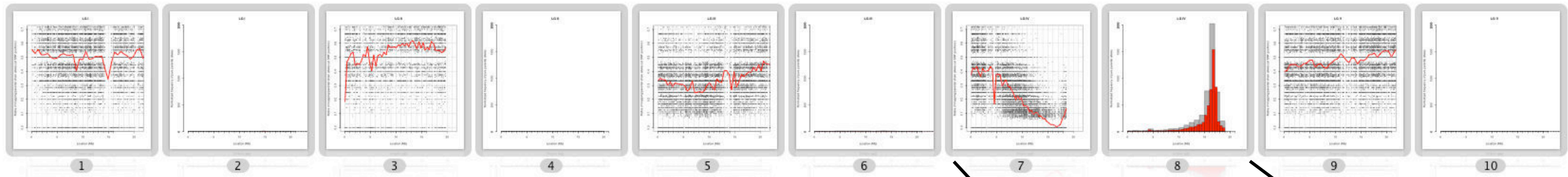
# Putative deletion analysis



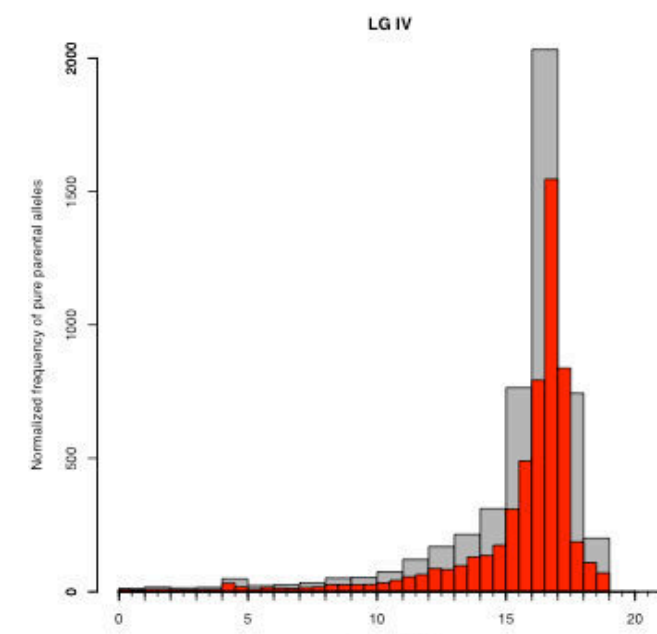
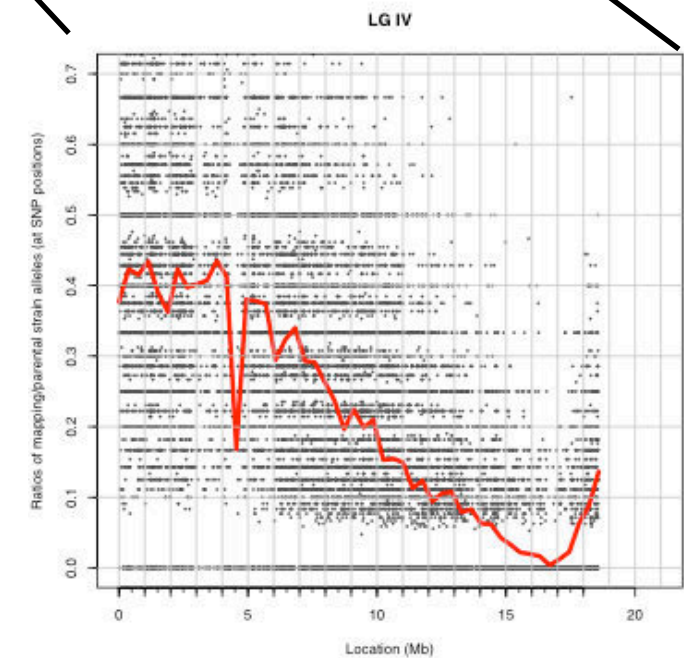
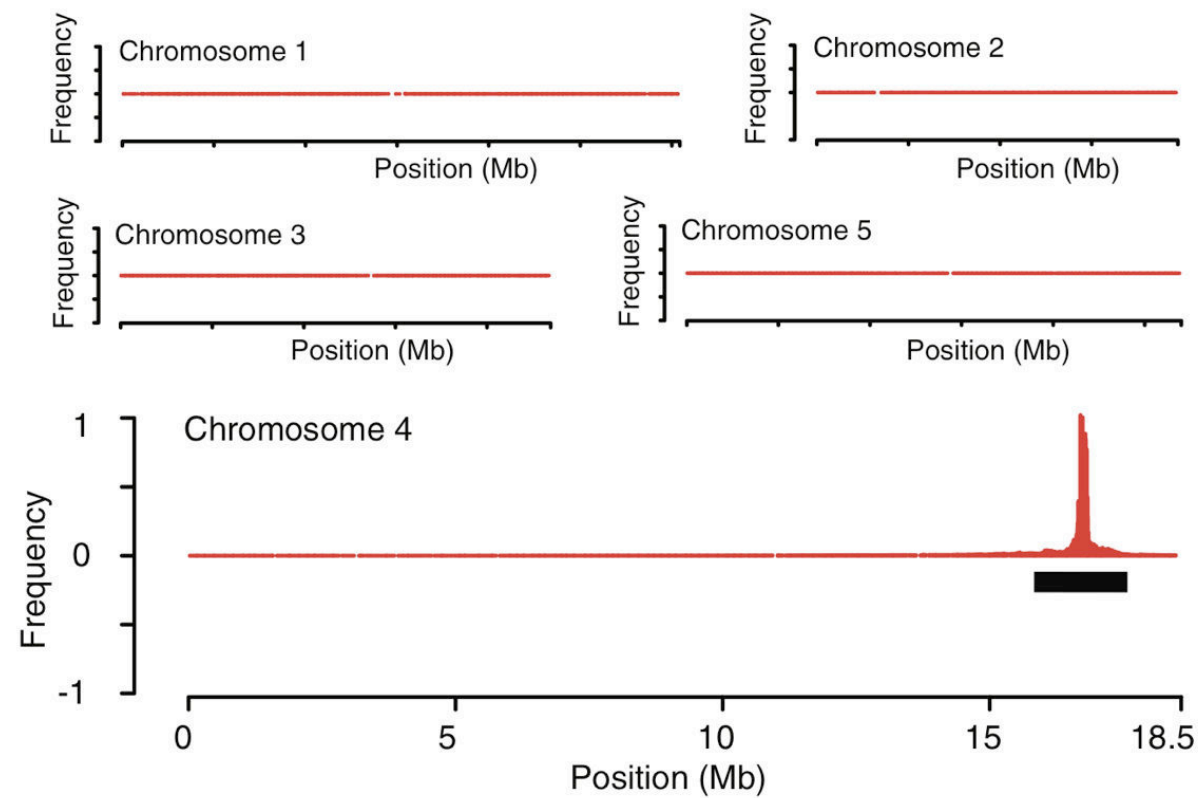


# Supports *Arabidopsis thaliana* (and other species)

Arabidopsis\_AT4G35090gene\_Galaxy39-[CloudMap\_SNP\_mapping\_with\_WGS].pdf



*y-axis: relative parental allele frequency*





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Daniel Blankenberg<sup>2</sup>, Anton Nekrutenko<sup>2</sup>, Oliver Hobert<sup>1</sup>

<sup>1</sup> Department of Biochemistry and Molecular Biophysics, HHMI  
Columbia University

<sup>2</sup> Center for Comparative Genomics and Bioinformatics,  
Penn State University

Special thanks to Jen Jackson, Nate Coraor and Dave  
Clements

<https://test.g2.bx.psu.edu/u/gal40/p/cloudmap>