Phase chrX



For each individual:
For each tissue:
Identify positions on chrX where
allele balance is equal to 1

### PT101159 heart

Allele balance

60624	3.0	13.0	0.23076923076923078
	5.0	13.0	0.230/09230/09230/8
694619	14.0	26.0	0.5384615384615384
879465	4.0	13.0	0.3076923076923077
1314201	19.0	26.0	0.7307692307692307
1436296	21.0	38.0	0.5526315789473685
1436503	22.0	40.0	0.55
1436515	22.0	38.0	0.5789473684210527
1717258	84.0	178.0	0.47191011235955055
1724510	9.0	46.0	0.1956521739130435
8720846	5.0	13.0	0.38461538461538464
	_		
Ī	Î		Total number of reads
<u>'</u>			
position			

Obtain the positions where the allele balance is equal to 1 (completely skewed)

15705225	172.0	172.0	1.0
79147400	26.0	26.0	1.0
79147429	30.0	30.0	1.0
147054400	29.0	29.0	1.0
147054504	29.0	29.0	1.0
147054527	24.0	24.0	1.0
147054535	27.0	27.0	1.0
147054564	24.0	24.0	1.0
147054617	21.0	21.0	1.0

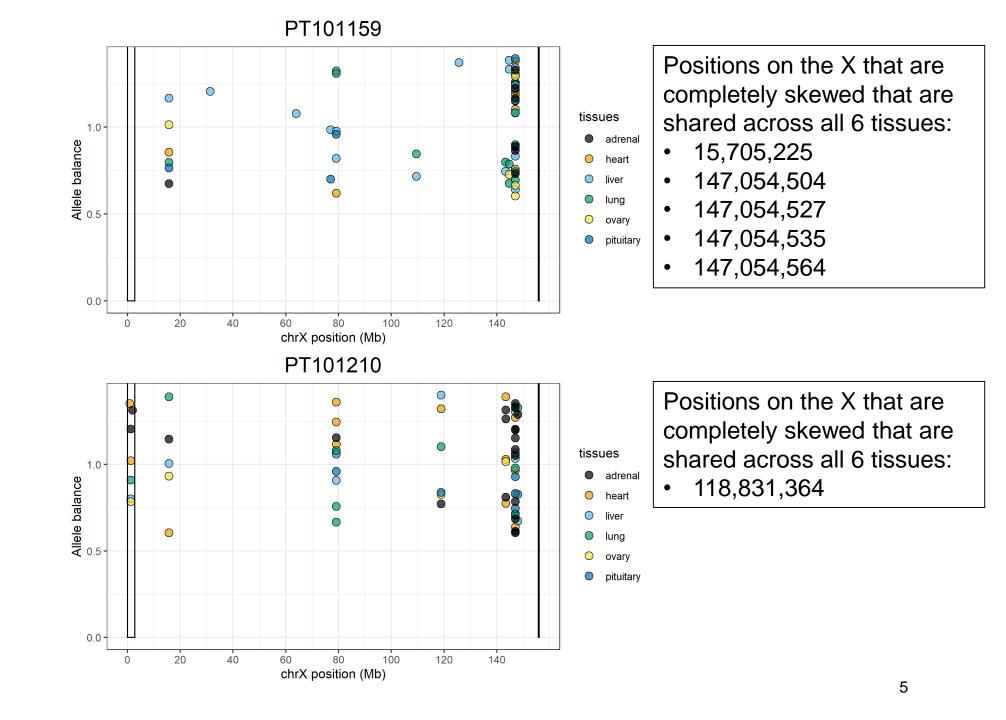
Number of reads for the biased allele (biased allele is determined from phasing

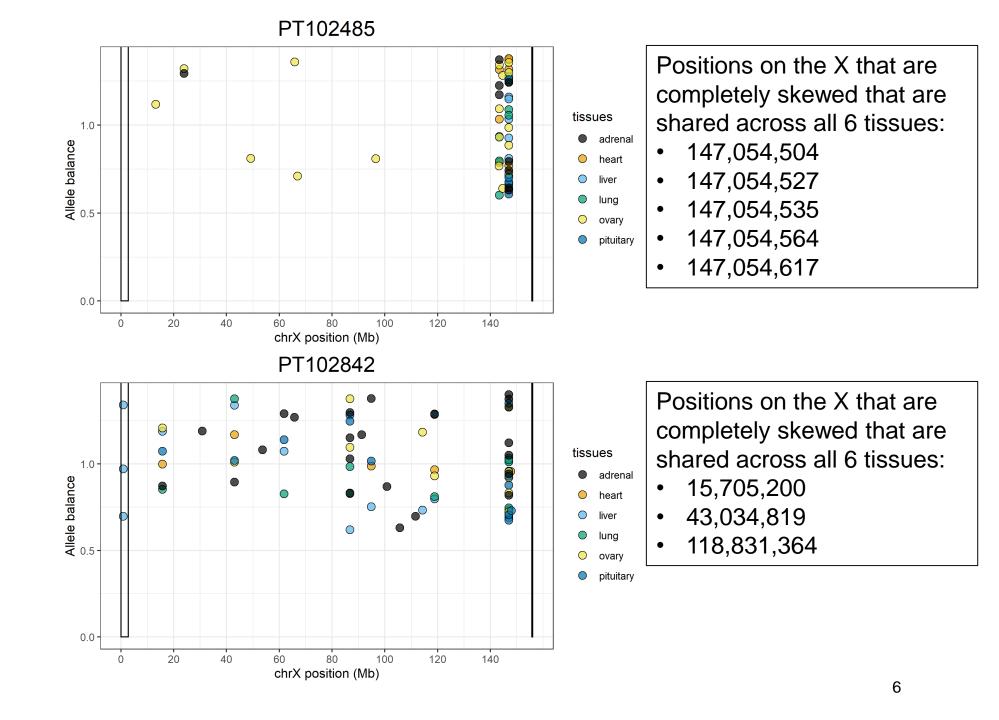


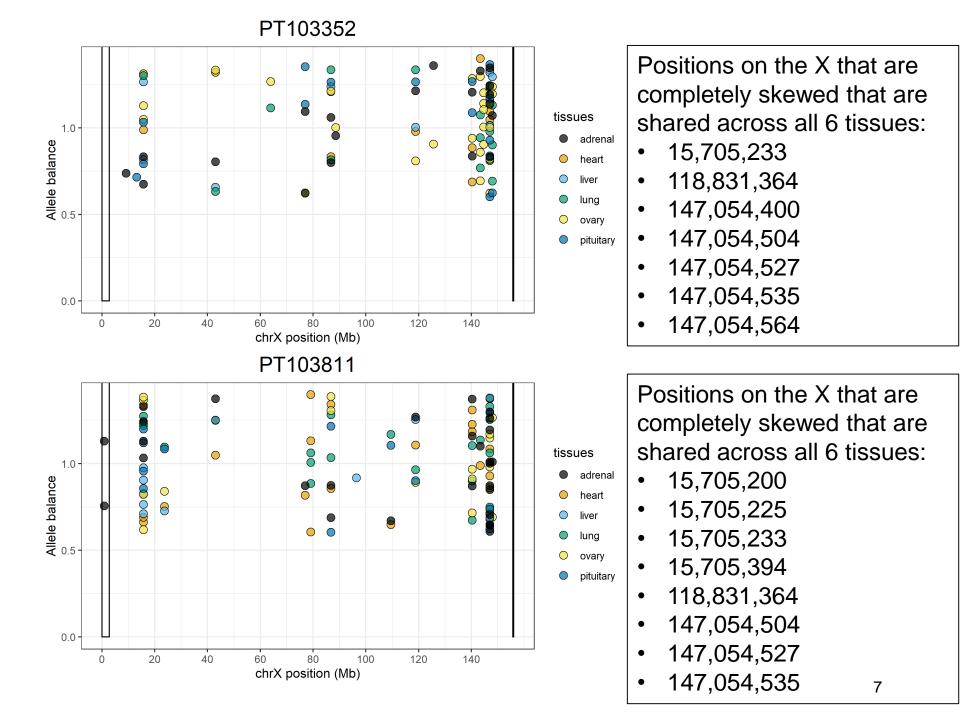
Repeat for all of the tissues within an individual

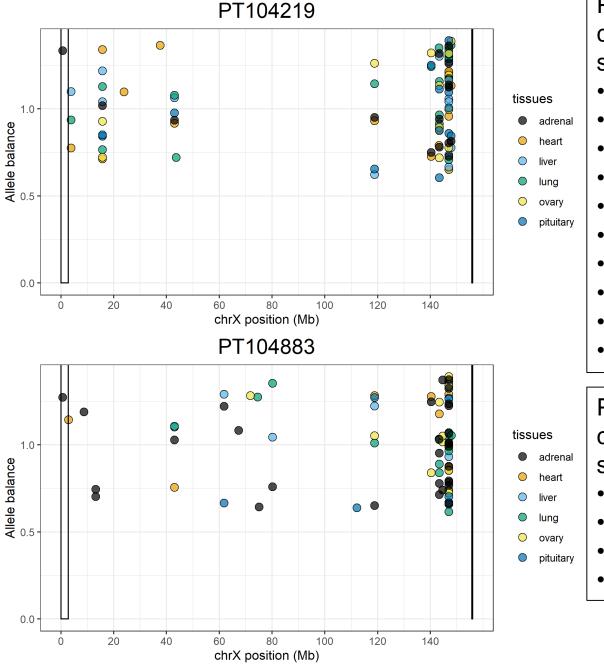
```
# PT101159
setwd("c://Users/tuyen/Documents/postdoc_asu/projects/Cayo_x_inactivation/whole_X_inactivation/results
/phase_whole_X/minDepth10/PT101159/")
heart = read.table("PT101159_hap1_heart_hardfilter_complete_skewed.tsv")
liver = read.table("PT101159_hap1_liver_hardfilter_complete_skewed.tsv")
lung = read.table("PT101159_hap1_lung_hardfilter_complete_skewed.tsv")
ovary = read.table("PT101159_hap1_gonads_hardfilter_complete_skewed.tsv")
pituitary = read.table("PT101159_hap1_pituitary_hardfilter_complete_skewed.tsv")
adrenal = read.table("PT101159_hap1_adrenal_hardfilter_complete_skewed.tsv")
heart_label = rep("heart", nrow(heart))
liver_label = rep("liver", nrow(liver))
lung_label = rep("lung", nrow(lung))
ovary_label = rep("ovary", nrow(ovary))
pituitary_label = rep("pituitary", nrow(pituitary))
adrenal_label = rep("adrenal", nrow(adrenal))
plot_data = data.frame(tissues=c(heart_label, liver_label, lung_label, ovary_label, pituitary_label,
adrenal_label), allele_balance = 1, positions = c(heart[,1], liver[,1], lung[,1], ovary[,1], pituitary[,1],
adrenal[,1]))
cbbPalette <- c("#000000", "#E69F00", "#56B4E9", "#009E73", "#F0E442", "#0072B2")
pnq("c://Users/tuyen/Documents/postdoc_asu/projects/Cayo_x_inactivation/whole_X_inactivation/results/phase_wh
ole_X/minDepth10/Plots/PT101159_complete_skewed.pnq", width = 7, height = 4, units = "in", res = 300)
qqplot(data=plot_data, aes(x=positions, y=allele_balance, fill=tissues)) +
  geom_point(shape=21, size=3, color="black", position="jitter")| +
 theme_bw() +
  coord_cartesian(xlim = c(1, 156040895)) +
  # geom_jitter(shape=21, size=4, color="black", aes(fill=tissues)) +
  scale_x_continuous(breaks=c(0, 20000000, 40000000, 60000000, 80000000, 100000000, 120000000, 140000000),
                     labels=c("0", "20", "40", "60", "80", "100", "120", "140")) +
  labs(x="chrx position (Mb)", y="Allele balance", title="PT101159") +
  theme(plot.title = element_text(hjust = 0.5, size=18)) +
  geom_rect(aes(xmin=10001, xmax=2781479, ymin=0, ymax=Inf), fill=NA, color="black") +
 qeom_rect(aes(xmin=155701383, xmax=156030895, ymin=0, ymax=Inf), fill=NA, color="black") +
 scale_fill_manual(values=alpha(cbbPalette, 0.7))
dev.off()
```

# Phased chrX Cayo







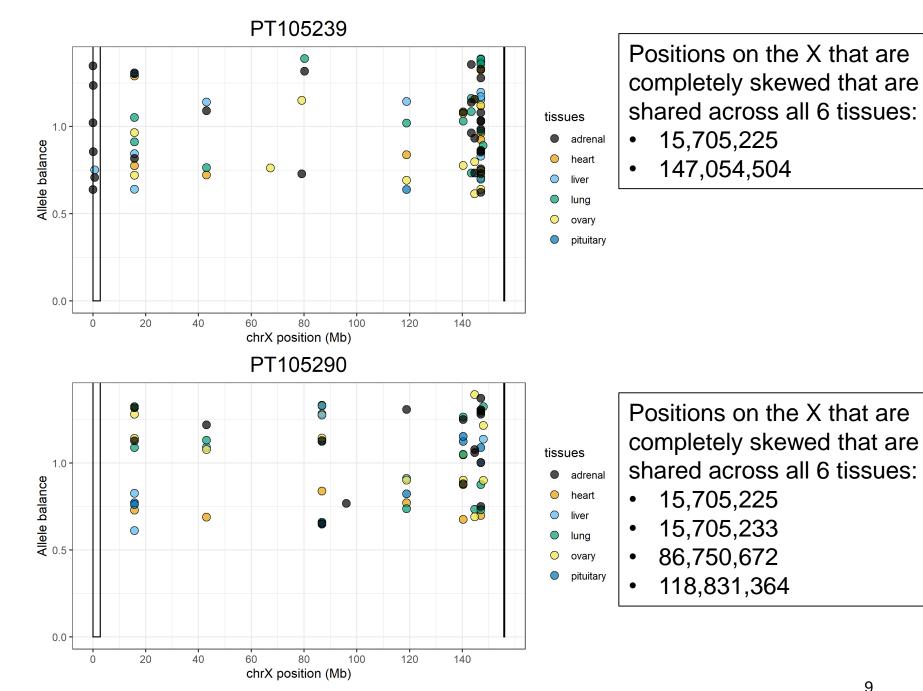


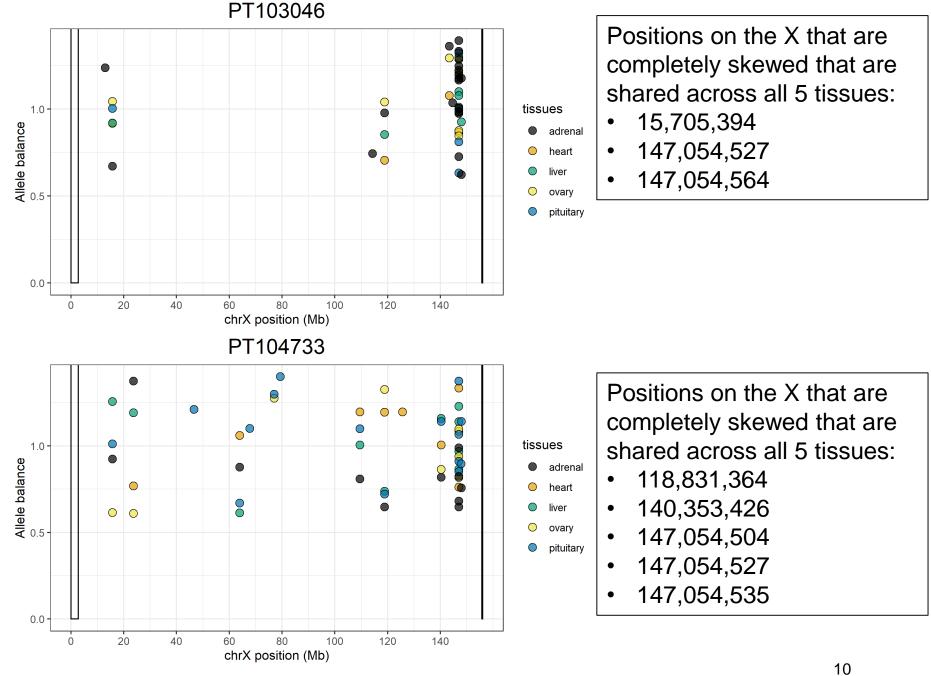
Positions on the X that are completely skewed that are shared across all 6 tissues:

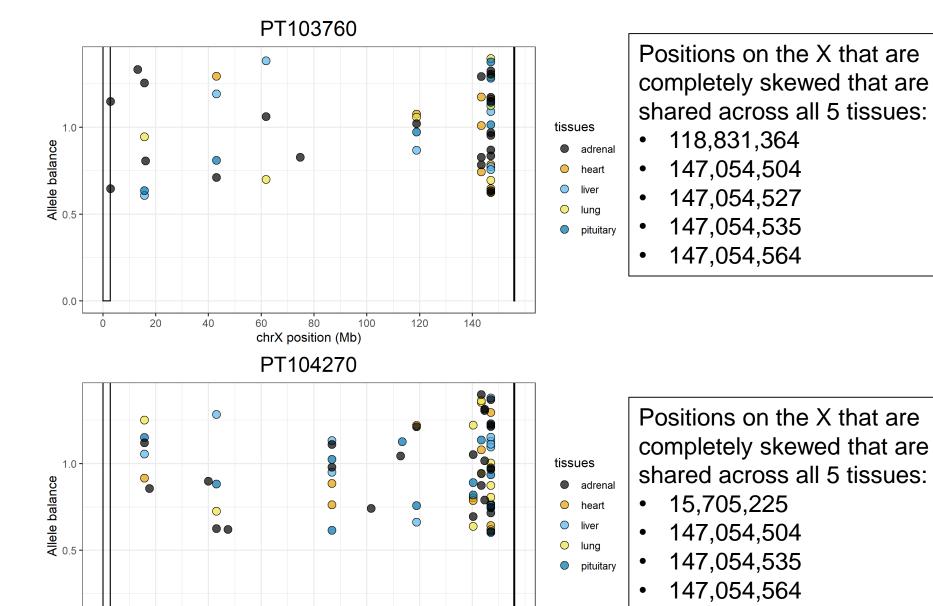
- 15,705,225
- 118,831,364
- 143,373,439
- 147,054,400
- 147,054,504
- 147,054,527
- 147,054,535
- 147,054,564
- 147,054,617
- 147,823,222

Positions on the X that are completely skewed that are shared across all 6 tissues:

- 118,831,364
- 147,054,527
- 147,054,535
- 147,054,564







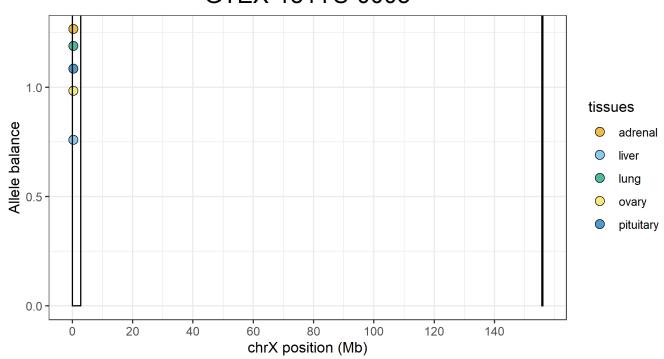
chrX position (Mb)

0.0

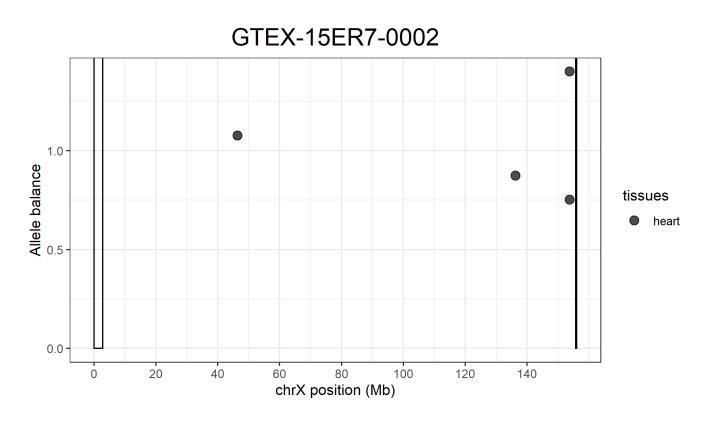
## Phased chrX GTEx

Tissue	How many variants that are completely skewed?	Positions
Heart	0	
Liver	1	311858
Lung	1	311858
Ovary	1	311858
Pituitary	1	311858
Adrenal	1	311858

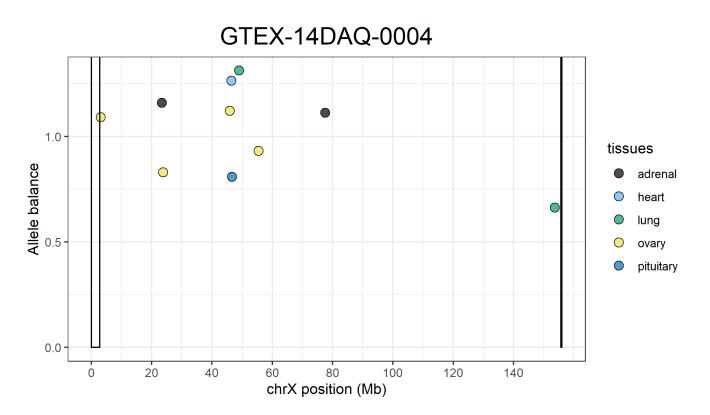




Tissue	How many variants that are completely skewed?	Positions
Heart	4	46440204, 136228626, 136228626, 153784707
Liver	0	
Lung	0	
Ovary	0	
Pituitary	0	
Adrenal	0	

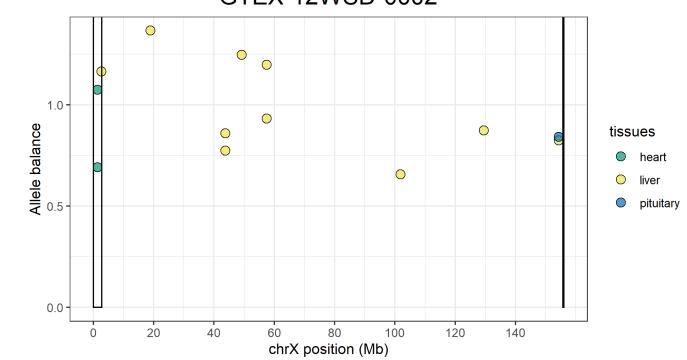


Tissue	How many variants that are completely skewed?	Positions
Heart	1	46440204
Liver	0	
Lung	2	48575111, 154462057
Ovary	4	3321504, 23783476, <b>46440204</b> , 54927214
Pituitary	1	46440204
Adrenal	2	23783476, 78125505

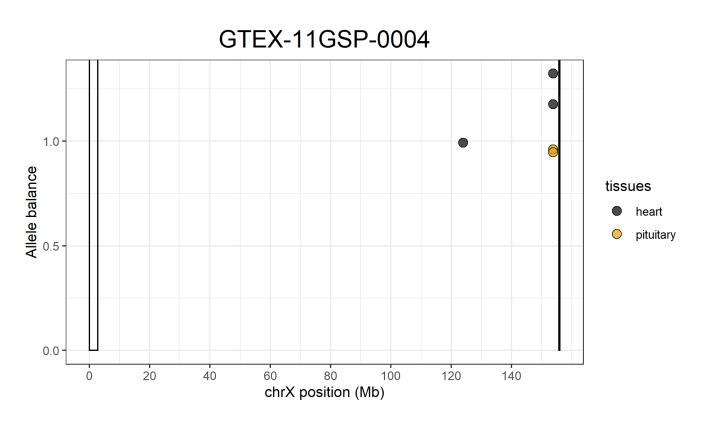


Tissue	How many variants that are completely skewed?	Positions
Heart	2	1412872, 1412877
Liver	10	2717448, 18893947, 43768752, 43768865, 49175859, 57448699, 57489006, 101886821, 129448500, <b>154348765</b>
Lung	0	
Ovary	0	
Pituitary	1	154348765
Adrenal	0	

### GTEX-12WSD-0002

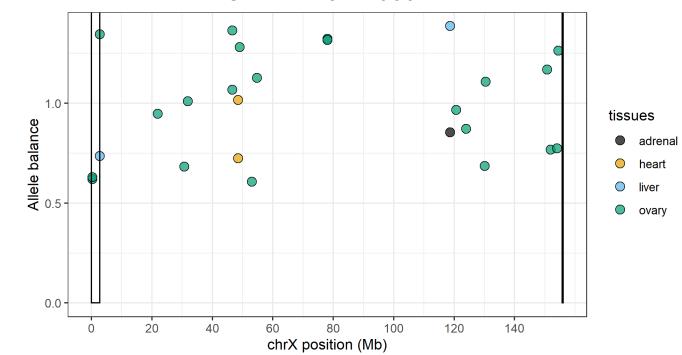


Tissue	How many variants that are completely skewed?	Positions
Heart	3	123913073, <b>153784705</b> , <b>153784707</b>
Liver	0	
Lung	0	
Ovary	0	
Pituitary	2	153784705, 153784707
Adrenal	0	



Tissue	How many variants that are completely skewed?	Positions
Heart	2	48577014, 48577017
Liver	2	2717448, <b>118685810</b>
Lung	0	
Ovary	21	284193, 290695, 2717448, 21967346, 30727673, 31875190, 46607325, 46613391, 49076231, 53087548, 54813580, 78125505, 78125517, 120626774, 123913432, 130067204, 130364862, 150762633, 151956016, 154014407, 154429242
Pituitary	0	
Adrenal	1	118685810

### GTEX-1A32A-0001



Tissue	How many variants that are completely skewed?	Positions
Heart	5	3818448, <b>10143330</b> , 46440204, 66207198, 153784707
Liver	1	48576920
Lung	2	10143330, 155309667
Ovary	2	10143330, 1 <u>55309667</u>
Pituitary	1	10143330
Adrenal	1	10143330

