

# Capstone\_Alignment

I used Tophat2 to do the alignment. The reference genome I used was hg19. Given the computational resources I have, I only used three adult samples and three fetal samples.

## Adult

### Adult\_1

#### Adult\_1\_Rep1 (SRR1554536)

Left reads:

Input : 21450348

Mapped : 21226856 (99.0% of input)

of these: 1211783 ( 5.7%) have multiple alignments (4044 have >20)

Right reads:

Input : 21450348

Mapped : 21236299 (99.0% of input)

of these: 1213596 ( 5.7%) have multiple alignments (4041 have >20)

99.0% overall read mapping rate.

Aligned pairs: 21029471

of these: 1198673 ( 5.7%) have multiple alignments

684409 ( 3.3%) are discordant alignments

94.8% concordant pair alignment rate.

#### Adult\_1\_Rep2 (SRR2071347)

Left reads:

Input : 37024538

Mapped : 32522655 (87.8% of input)

of these: 1818097 ( 5.6%) have multiple alignments (5773 have >20)

Right reads:

Input : 37024538

Mapped : 30805216 (83.2% of input)

of these: 1680278 ( 5.5%) have multiple alignments (5766 have >20)

85.5% overall read mapping rate.

Aligned pairs: 28924189

of these: 1521568 ( 5.3%) have multiple alignments

849826 ( 2.9%) are discordant alignments

75.8% concordant pair alignment rate.

### Adult\_2

#### Adult\_2\_Rep1 (SRR1554539)

Left reads:

Input : 33742728

Mapped : 33165728 (98.3% of input)  
of these: 1251939 ( 3.8%) have multiple alignments (12991 have >20)

Right reads:

Input : 33742728

Mapped : 33088048 (98.1% of input)

of these: 1248235 ( 3.8%) have multiple alignments (12989 have >20)

98.2% overall read mapping rate.

Aligned pairs: 32573446

of these: 1223711 ( 3.8%) have multiple alignments

745730 ( 2.3%) are discordant alignments

94.3% concordant pair alignment rate.

### **Adult\_\_2\_Rep2 (SRR2071350)**

Left reads:

Input : 49643890

Mapped : 39200576 (79.0% of input)

of these: 1471665 ( 3.8%) have multiple alignments (13146 have >20)

Right reads:

Input : 49643890

Mapped : 36196122 (72.9% of input)

of these: 1349243 ( 3.7%) have multiple alignments (13137 have >20)

75.9% overall read mapping rate.

Aligned pairs: 32579223

of these: 1197551 ( 3.7%) have multiple alignments

742978 ( 2.3%) are discordant alignments

64.1% concordant pair alignment rate.

### **Adult\_\_3**

#### **Adult\_\_3\_Rep1 (SRR1554534)**

Left reads:

Input : 28181772

Mapped : 27604689 (98.0% of input)

of these: 1192906 ( 4.3%) have multiple alignments (7067 have >20)

Right reads:

Input : 28181772

Mapped : 27493559 (97.6% of input)

of these: 1186717 ( 4.3%) have multiple alignments (7066 have >20)

97.8% overall read mapping rate.

Aligned pairs: 26995064

of these: 1159712 ( 4.3%) have multiple alignments

516215 ( 1.9%) are discordant alignments

94.0% concordant pair alignment rate.

#### **Adult\_\_3\_Rep2 (SRR2071345)**

Left reads:

Input : 41066237

Mapped : 32031158 (78.0% of input)  
of these: 1369584 ( 4.3%) have multiple alignments (7203 have >20)

Right reads:

Input : 41066237

Mapped : 29882532 (72.8% of input)

of these: 1269949 ( 4.2%) have multiple alignments (7184 have >20)

75.4% overall read mapping rate.

Aligned pairs: 27008790

of these: 1133370 ( 4.2%) have multiple alignments

492619 ( 1.8%) are discordant alignments

64.6% concordant pair alignment rate.

## **Fetal**

### **Fetal\_\_1**

#### **Fetal\_\_1\_\_Rep1 (SRR1554537)**

Left reads:

Input : 55133946

Mapped : 53971223 (97.9% of input)

of these: 2650409 ( 4.9%) have multiple alignments (38477 have >20)

Right reads:

Input : 55133946

Mapped : 54295543 (98.5% of input)

of these: 2668079 ( 4.9%) have multiple alignments (38486 have >20)

98.2% overall read mapping rate.

Aligned pairs: 53240197

of these: 2593284 ( 4.9%) have multiple alignments

1431670 ( 2.7%) are discordant alignments

94.0% concordant pair alignment rate.

#### **Fetal\_\_1\_\_Rep2 (SRR2071348)**

Left reads:

Input : 125126837

Mapped : 105156259 (84.0% of input)

of these: 5147120 ( 4.9%) have multiple alignments (62885 have >20)

Right reads:

Input : 125126837

Mapped : 98624858 (78.8% of input)

of these: 4788256 ( 4.9%) have multiple alignments (62712 have >20)

81.4% overall read mapping rate.

Aligned pairs: 90687653

of these: 4344495 ( 4.8%) have multiple alignments

2352198 ( 2.6%) are discordant alignments

70.6% concordant pair alignment rate.

## **Fetal\_2**

### **Fetal\_2\_Rep1 (SRR1554567)**

Left reads:

Input : 61922935

Mapped : 61063039 (98.6% of input)

of these: 2588779 ( 4.2%) have multiple alignments (37226 have >20)

Right reads:

Input : 61922935

Mapped : 60764550 (98.1% of input)

of these: 2569329 ( 4.2%) have multiple alignments (37218 have >20)

98.4% overall read mapping rate.

Aligned pairs: 60018416

of these: 2515340 ( 4.2%) have multiple alignments

1291310 ( 2.2%) are discordant alignments

94.8% concordant pair alignment rate.

### **Fetal\_2\_Rep2 (SRR2071378)**

Left reads:

Input : 77669693

Mapped : 70304205 (90.5% of input)

of these: 3021579 ( 4.3%) have multiple alignments (37550 have >20)

Right reads:

Input : 77669693

Mapped : 63645698 (81.9% of input)

of these: 2681403 ( 4.2%) have multiple alignments (37456 have >20)

86.2% overall read mapping rate.

Aligned pairs: 60038624

of these: 2492847 ( 4.2%) have multiple alignments

1294283 ( 2.2%) are discordant alignments 75.6% concordant pair alignment rate.

## **Fetal\_3**

### **Fetal\_3\_Rep1 (SRR1554568)**

Left reads:

Input : 48184702

Mapped : 47468224 (98.5% of input)

of these: 2033100 ( 4.3%) have multiple alignments (28182 have >20)

Right reads:

Input : 48184702

Mapped : 47308913 (98.2% of input)

of these: 2021478 ( 4.3%) have multiple alignments (28201 have >20)

98.3% overall read mapping rate.

Aligned pairs: 46694237

of these: 1977088 ( 4.2%) have multiple alignments

973031 ( 2.1%) are discordant alignments

94.9% concordant pair alignment rate.

### **Fetal\_3\_Rep2 (SRR2071379)**

Left reads:

Input : 119227332

Mapped : 102136596 (85.7% of input)

of these: 4523322 ( 4.4%) have multiple alignments (58069 have >20)

Right reads:

Input : 119227332

Mapped : 95094126 (79.8% of input)

of these: 4169064 ( 4.4%) have multiple alignments (57918 have >20)

82.7% overall read mapping rate.

Aligned pairs: 87885156

of these: 3795764 ( 4.3%) have multiple alignments

1914803 ( 2.2%) are discordant alignments

72.1% concordant pair alignment rate.