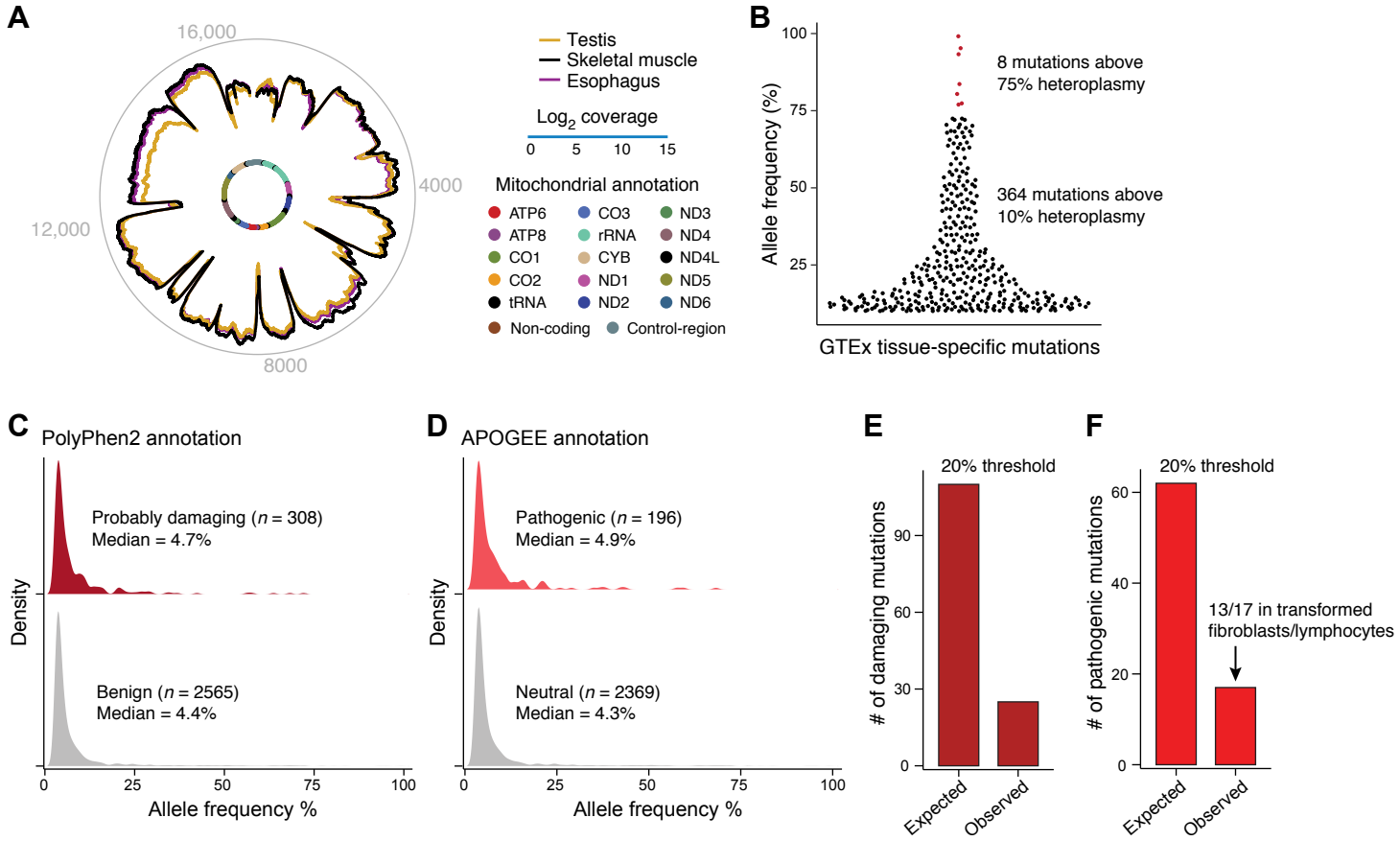
# Figure S4


A
16,000
4000
8000
12,000
Testis
Skeletal muscle
Esophagus
Log2 coverage
0 5 10 15
Mitochondrial annotation
ATP6
ATP8
CO1
CO2
tRNA
CO3
rRNA
CYB
ND1
ND2
ND3
ND4
ND4L
ND5
ND6
Non-coding
Control-region
B
Allele frequency (%)
100
75
50
25
8 mutations above
75% heteroplasmy
364 mutations above
10% heteroplasmy
GTEx tissue-specific mutations
C
PolyPhen2 annotation
Density
Probably damaging (n = 308)
Median = 4.7%
Benign (n = 2565)
Median = 4.4%
0 25 50 75 100
Allele frequency %
D
APOGEE annotation
Density
Pathogenic (n = 196)
Median = 4.9%
Neutral (n = 2369)
Median = 4.3%
0 25 50 75 100
Allele frequency %
E
20% threshold
# of damaging mutations
0 30 60 90
Expected
Observed
F
20% threshold
# of pathogenic mutations
0 20 40 60
13/17 in transformed
fibroblasts/lymphocytes
Expected
Observed