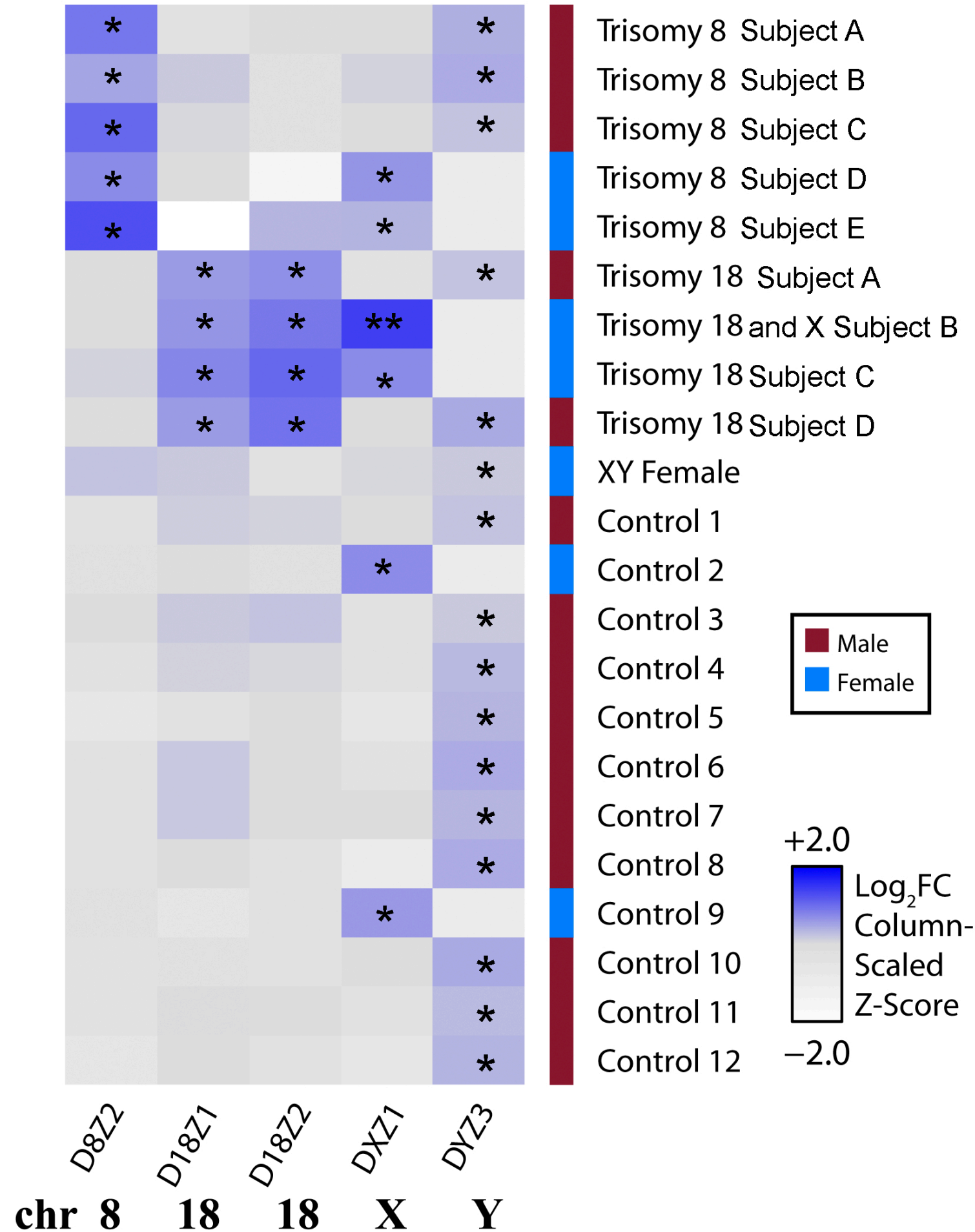
Trisomy 8 Subject A
Trisomy 8 Subject B
Trisomy 8 Subject C
Trisomy 8 Subject D
Trisomy 8 Subject E
Trisomy 18 Subject A
Trisomy 18 and X Subject B
Trisomy 18 Subject C
Trisomy 18 Subject D
XY Female
Control 1
Control 2
Control 3
Control 4
Control 5
Control 6
Control 7
Control 8
Control 9
Control 10
Control 11
Control 12
Male
Female
+2.0
Log2FC Column-Scaled Z-Score
−2.0
D8Z2
D18Z1
D18Z2
DXZ1
DYZ3
chr 8 18 18 X Y

Figure S9. Variations in the content of centromere repeats in somatic and sex chromosomes identify ploidy. Heat map representing the abundance of the somatic centromere arrays D8Z2, D18Z1, and D18Z2 (in chromosomes 8 and 18, respectively) and the α-repeat arrays in the sex chromosomes, DXZ1 and DYZ3 (X axis), in DNA from individuals of different gender, including individuals with somatic trisomy 8 and 18 (Y axis). The phenotypic gender of the individuals is represented by brown or blue color bars on the right, with brown representing males and blue representing females. The gradient bar on the right indicates the color intensity of the heat map, with overrepresented repeats in blue and underrepresented repeats in white. A star indicates significant differences in the content of α-repeats; as expected, all subjects have at least one sex chromosome that is highly represented. Trisomy 18/X Subject B is an individual with trisomies 18 and X. The XY female is a subject who is phenotypically female but genetically male. As expected, DYZ3 from chromosome Y was detected only in the male population (blue bands with stars), except for the XY female. The number of α-repeats of the chromosome X DXZ1 was higher in females than in males and much higher in an individual with trisomies 18 and X. Individuals with trisomy 8 have significantly higher content of D8Z2 repeats, whereas individuals with trisomy 18 have significantly higher content of D18Z1 and D18Z2 repeats. Thus, the centromeric PCR assays quickly and accurately detect ploidy for these chromosomes. Statistical differences between the size of centromere arrays in chr 8 and 18 between patients with trisomy 8, trisomy 18, and healthy individuals were calculated using the ANOVA test and significant differences between the groups estimated using the Dunnett's multiple comparisons test. Two-tailed p values were considered significant at $p < 0.05$.