

Fig 3. Comparison between male and female pregnancies and aneuploidy samples

The performance of the assay was evaluated using 123 samples including 51 female and 58 male euploid samples and 14 trisomy 21 samples. The performance between male (n=51) and female (n=59) pregnancy samples did not differ (p-value=0.97; Wilcoxon test). The fetal fraction estimation accuracy was further assessed on 14 trisomy 21 samples showing comparable performance to the euploid male samples (p-value=0.13; Wilcoxon test). The maternal background (noise) level at very low fetal fractions was assessed using 8 non-pregnant samples. The estimated fetal fractions ranged from 0.2% to 1.09%.