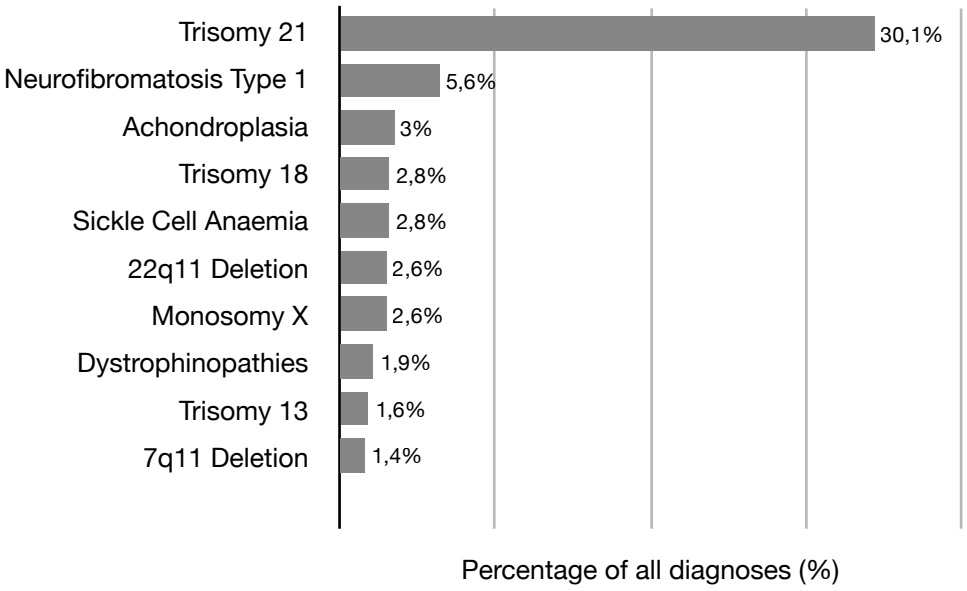
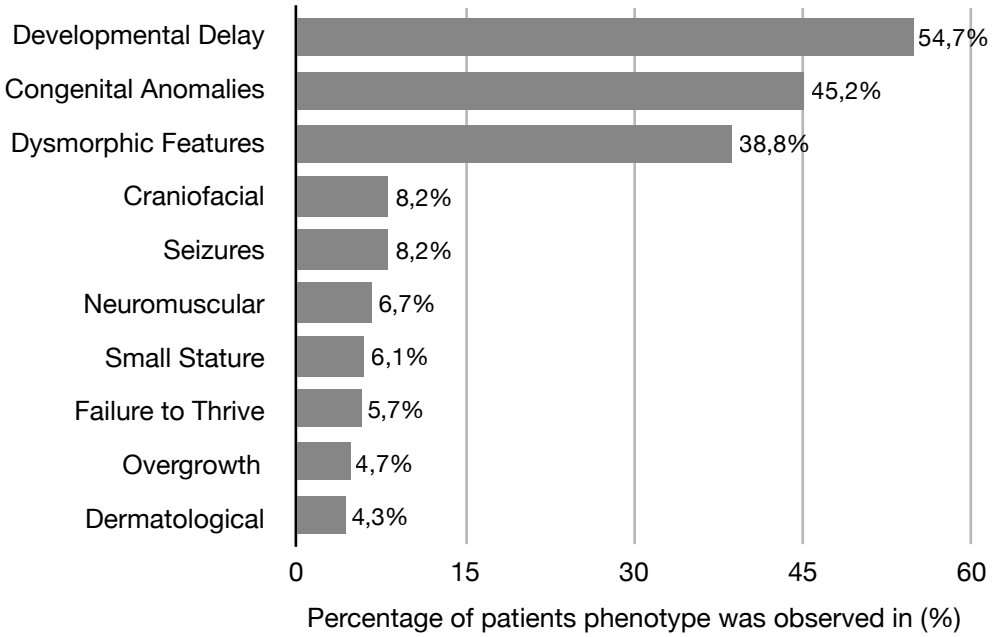
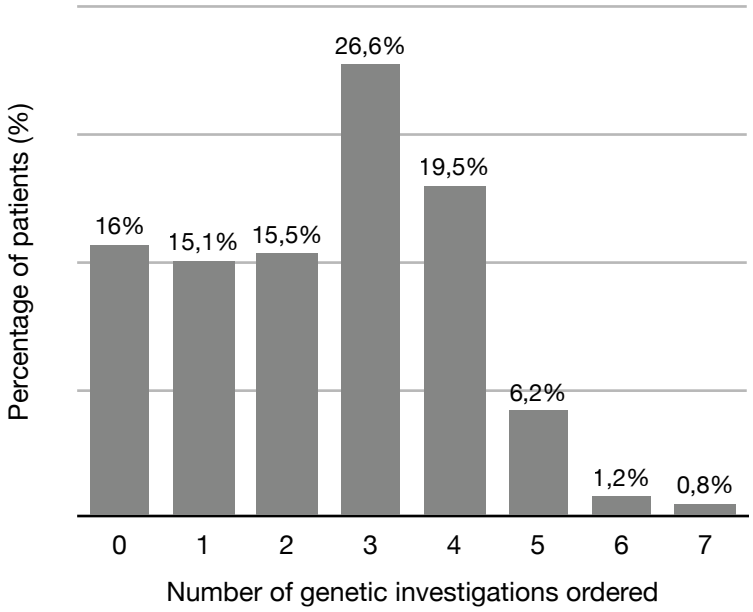
Supplementary Figures



Supplementary Figure 1: Top 10 genetic diagnoses made in the cohort. These top ten diagnoses account for 54% of all diagnoses, with Trisomy 21 being the most common diagnosis made.



Supplementary Figure 2: Top ten phenotypes observed in patients in Group C The most common phenotypes by far were developmental delay, congenital anomalies and dysmorphic features.



Supplementary Figure 3: Number of genetic investigations undergone by undiagnosed patients in Group C. The greatest percentage (26,6%) of patients had 3 tests ordered and 54.3% of patients had 3 or more tests.