

[Estimate of the contemporary live-birth prevalence of recurrent 22q11.2 deletions: a cross-sectional analysis from population-based newborn screening \[Full text\]](#)

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In this study, our team screened for 22q deletions in just over 30,000 babies born in Ontario between January 2017 and September 2018, in order to determine the live birth prevalence of 22q. We found 14 newborns with confirmed 22q deletions in the sample, which corresponds to an estimated live birth prevalence of 1 in 2148 live births. To put this number into perspective, cystic fibrosis and severe combined immunodeficiency (two of the genetic conditions that are screened for in Ontario's existing Newborn Screening program) in fact have lower prevalence rates than that of 22q. We also found that the babies with 22q likely to have younger mothers, were smaller in size for their gestational age, and had lower TREC levels, which can be a marker for immunodeficiency. These results support the importance of early 22q diagnosis (either prenatally or in infancy through newborn screening), which would allow for earlier screening and management of features associated with 22q.