Increased Nuchal Translucency and Developmental Delay in Fetuses with Normal Karyotype and Normal Anatomy: A Systematic Review and Meta-analysis

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Objective: To pool published data regarding the association of increased nuchal translucency (NT) with neurodevelopmental delay in fetuses with normal karyotype and normal anatomy.

Methods: Literature search of PubMed, Web of Sciences, Scopus, and Cochrane Library were performed from their inception to January 2022 to identify eligible studies on increased NT with neurodevelopmental outcome. The search included the combination of terms "nuchal translucency" and "outcome". Only fetuses with increased nuchal translucency with normal karyotype and no structural defects or syndromic abnormalities were analyzed. Between-studies heterogeneity was assessed using the I^2 statistic.

Result: The total prevalence of neurodevelopmental delay in 25 studies was 356/9274 (3.8%). Eleven studies (n=3100) used NT>99th centile as the cut-off; 58 children (1.8%; 95% CI, 0.001-0.036) were evaluated as having neurodevelopmental delay (I^2=85.1%). Nine studies (n=6461) used the 95th centile as the cut-off; 275 children were diagnosed with neurodevelopmental delay (4.2%; 95% CI, 0.012-0.041; I^2=81.8).

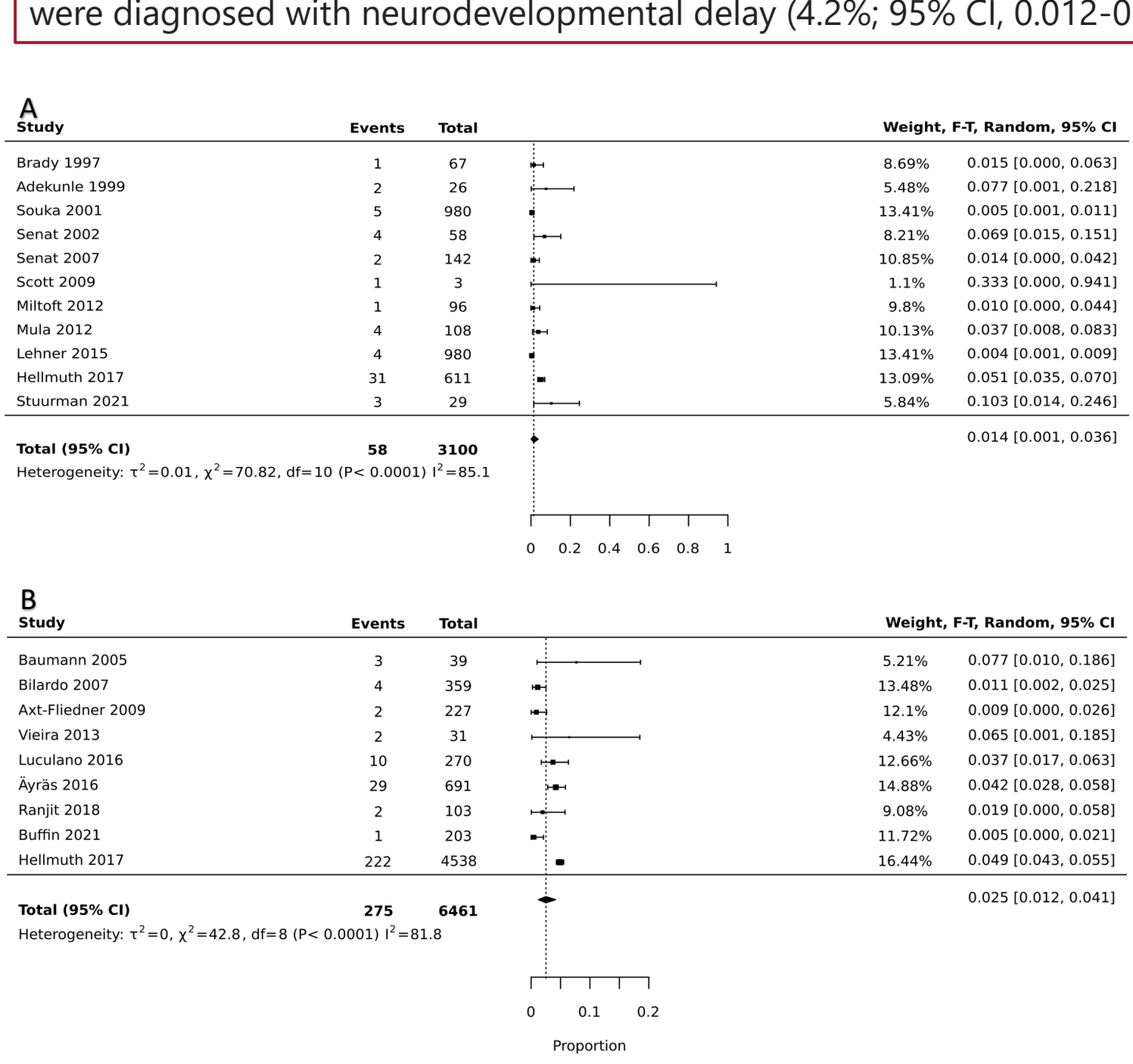

Figure 1: Forest plots of neurodevelopmental delay in children with increased NT. Studies that used NT>99th centile as cut-off (A) and studies that used NT>95th centile as cut-off (B).

Conclusion: The rate of neurodevelopmental delay in children with increased fetal NT, a normal karyotype and normal structure seems to be higher than the general population. However, there is high heterogeneity across studies, which largely remains even in subgroup analysis of studies of apperantly similar design potentially indicating the presence of some residual unidentified bias.