Prevalence of major and minor congenital anomalies in a cohort of children with fetal alcohol spectrum disorder (FASD): A national study

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Introduction: Children with prenatal alcohol exposure in the first trimester are at an increased risk of congenital anomalies, including fetal alcohol spectrum disorder (FASD), which is classified as a major congenital anomaly. There have been no nation-wide studies of congenital anomalies occurring in children with FASD in Australia. This study examined the range and prevalence of major and minor congenital anomalies in a national cohort of children with FASD.

Method: Active prospective case ascertainment was performed by the Australian Paediatric Surveillance Unit with monthly case reporting by paediatricians of newly diagnosed cases of FASD in children aged under 15 years [1]. Reports were made between January 2015 through April 2024. Reported congenital anomalies were classified based on standardised classification systems (ICD-10 and WARDA), and prevalence rates were compared with available population and control cohort rates.

Key Findings: Ninety paediatricians reported 1230 cases of FASD fulfilling criteria, 988 of which had ≥1 congenital anomaly in addition to FASD. There were 152 additional anomalies identified, with a median of 3 anomalies per child, including FASD (range: 1 to 14). The most prevalent major anomaly after FASD was microcephaly (20.5%) and was significantly more prevalent than in the population ($p < 0.0001$). The most prevalent minor anomalies were the sentinel facial features of FASD (short palpebral fissures [54.2%]; smooth philtrum [66.3%]; thin upper lip [60.1%]), aligning with previous findings [2], followed by clinodactyly (19.8%).

Discussions and Conclusions: This is the first nation-wide study of congenital anomalies in children with FASD in Australia. Cases were reported based on strict diagnostic criteria with reporting in real time by specialist paediatricians. The range and prevalence of congenital anomalies reported demonstrates the extensive impacts of prenatal alcohol exposure in the first trimester. More comparative data from other FASD cohorts, control cohorts, and general populations is urgently required.

Implications for Practice or Policy: Increased recognition and documentation of additional congenital anomalies by clinicians may allow for earlier identification of cases of FASD. For the public, this report highlights the necessity to avoid alcohol when planning and in the early stages of pregnancy.

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