

Descriptive epidemiology of holoprosencephaly and arhinencephaly in metropolitan Atlanta, 1968-1992. ^[1]

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Abstract

We report the descriptive epidemiology of holoprosencephaly and arhinencephaly using data from the Metropolitan Atlanta Congenital Defects Program, a population-based birth defects surveillance system with multiple sources of ascertainment. From 1968-1992, we ascertained 63 cases of holoprosencephaly and arhinencephaly from approximately 734,000 births, for a birth prevalence of 0.86 per 10,000. Thirteen case infants with holoprosencephaly and four case infants with arhinencephaly were categorized as having syndromes. Of the case infants with non-syndromic holoprosencephaly, 55% had malformations not attributable to the underlying brain defect. The rate of holoprosencephaly and arhinencephaly increased from 0.58 per 10,000 during 1968-1972 to 1.2 per 10,000 during 1988-1992 (P for trend = 0.016). Rates were higher for females than for males (risk ratio = 1.45, 95% C.I. 0.88-2.41) and higher for nonwhites than for whites (risk ratio = 1.74, 95% C.I. 1.06-2.86). There was a U-shaped distribution of risk associated with maternal age with a slightly increased risk for younger women (risk ratio for maternal age < 20 years, compared with age 25-29 years = 1.68, 95% C.I. 0.77-3.62) and older women (risk ratio for maternal age > 34 years, compared with age 25-29 years = 2.30, 95% C.I. 0.93-5.7), but this was not statistically significant. The increased risk in the older age group could be largely explained by the presence of cases with autosomal trisomies. Neonatal mortality was higher for infants with malformations that were not attributable to the underlying brain defect and for infants with syndromes than for infants with isolated holoprosencephaly. This analysis is the first population-based study with long-term data on this rare defect. Further epidemiologic studies will be necessary to assess the risk factors for holoprosencephaly and arhinencephaly.

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