Changing demography of trisomy 18

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SUMMARY The incidence of trisomy 18 in Leicestershire during the years 1980–85 inclusive was one in 3086 births. Eleven of the 21 babies born with trisomy 18 in this period were delivered by caesarean section. Median and mean periods of survival were 2.5 and 22 days, respectively.

Parents who have had a child with a serious and life threatening disorder such as trisomy 18 (Edwards’ syndrome) often seek information concerning its local demography and likely prognosis. Experience of trisomy 18 in Leicestershire over the last six years suggests that this disorder is more common than generally recognised and that the pattern of survival is changing.

Patients and methods

Data for the years 1980–85 inclusive concerning all cases of trisomy 18 delivered in Leicestershire plus those terminated after diagnosis at amniocentesis were obtained from the records of (1) the Leicestershire perinatal mortality survey, (2) all local neonatal units, (3) the clinical genetics service, and (4) the cytogenetics units in Leicester, Nottingham, and Sheffield. The maternal and paediatric records of all cases were obtained and family visits were conducted after approval by the local ethical committee to obtain full details of family pedigrees.

Results

Incidence. Twenty three cases were ascertained (nine male and 14 female), of which two were terminated after diagnosis at amniocentesis. When these two cases are included the incidence during the study (total births 70 985) is one in 3086 births.

Obstetric data. Hydramnios was noted during the third trimester in 11 of the 21 pregnancies. Mean gestation at delivery was 36.7 weeks. Eleven of the 21 babies were delivered by caesarean section, electively in four cases because of intrauterine growth retardation and poor placental function and
by emergency in the remaining seven because of fetal distress during labour.

Clinical and genetic data. Mean weight, length, and head circumference at birth were 1580 g, 41.9 cm, and 30.4 cm, respectively. Mean (SD) parental ages at birth were 30.02 (7.63) years for fathers (mean maternal age for all births in England and Wales in 1982=29.63 years; t=0.245, p=0.5) and 28.56 years for mothers (mean maternal age for all births in England and Wales in 1982=26.37 years; t=1.32, p=0.1). Six mothers were aged over 35 years at the time of delivery.

In each case the diagnosis was suspected at or shortly after birth and was confirmed as full trisomy 18 by conventional G banded karyotype. Median and mean survival were 2.5 and 22 days, respectively. The range of survival was from 45 minutes to 18 weeks. One third of the babies died within 24 hours, and two thirds died within the first five days (Figure). Necropsy was performed in nine cases, revealing congenital cardiac abnormality in eight and tracheo-oesophageal fistula with oesophageal atresia in four.

Discussion

This retrospective survey of trisomy 18 yielded three unexpected observations.

Firstly, the incidence of 1 in 3086 births was higher than that noted in surveys of comparable size. An early study of the incidence of trisomy 18 found three cases in a consecutive series of 10,345 births occurring in a hospital in Wisconsin, giving an incidence of roughly 1 in 3500.1 Larger surveys have suggested lower incidence figures of around 1 in 5000 to 1 in 7000 births.2 3 We have no explanation for the higher incidence noted in Leicestershire, which showed no clear temporal, geographical, or social class clustering.

Secondly, the incidence of delivery by caesarean section, which was performed in 11 of the 21 cases, was high. This confirms a previous report4 and illustrates the potential value of developing a safe means for rapid fetal karyotyping during the third trimester for cases in which a severe fetal abnormality is suspected because of intrauterine growth retardation or hydramnios, or both.

Finally, the data derived in this study indicate that the pattern of survival in trisomy 18 is changing. It was noted 20 years ago that over 80% of trisomy 18 babies survived for at least two weeks, with 50% surviving for at least two months.5 A more recent study of cases born during the years 1972–82 in Queensland yielded median and mean life expectancies of five and 48 days respectively,2 compared with values of 2.5 and 22 days obtained in this study. This tendency towards shorter survival may be explained by changing patterns of patient management associated with early diagnosis. For those involved in counselling parents of a neonate with trisomy 18 it is important to remember that survival into childhood and beyond has been noted on rare occasions.6

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