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Short communication

Spontaneous pregnancy outcome after prenatal diagnosis of anencephaly

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Parents are usually told that many anencephalic offspring die *in utero* or soon after delivery, and many obstetricians offer elective termination of the pregnancy. Following the personal experience of the first author, a personal website was created with the intention of providing information and exchanging views with other parents confronted with a prenatal diagnosis of anencephaly. Data were collected from 211 pregnancies where the parents opted not to terminate pregnancy. These data revealed that polyhydramnios was a feature in 56 (26%) pregnancies, death *in utero* in 15 (7%) pregnancies, 72 (34%) babies were born prematurely (<37 weeks of gestation), 113 (53%) at term and 21 (10%) after 42 weeks. Stillbirth, presumably resulting from intrapartum death, occurred in 43 (20%) deliveries. One hundred and fifty-three (72%) of anencephalic offspring were liveborn, of those, 103 (67%) died within 24 hours but 6/211 survived 6 or more days (maximum 28 days). Continuation of pregnancy after a diagnosis of anencephaly is medically safe and should be considered as an option.

Keywords Anencephaly, prenatal diagnosis, outcome.

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Introduction

In recent years, neural tube defects (NTD) have attracted increased attention.[1] One important reason is the observation that the majority of NTD are preventable by appropriate folic acid supplementation.[2–4] Progress in epidemiological studies, as well as observation in animal models, suggests that the various types of NTD (i.e. anencephaly, spina bifida or myelomeningocele and closed spinal dysraphisms) differ in their pathogenesis.[5,6] Anencephaly represents the most severe form of NTD, uniformly lethal in the neonatal period, and it is one of the most lethal among congenital defects.[7]

The introduction of prenatal ultrasonography and maternal serum alphafetoprotein screening has resulted in prenatal detection of anencephaly in virtually 100% of cases.[8] This prompted obstetricians and parents to consider elective pregnancy termination. It appears that this option became routine in the 1990s, and in many centres, the birth rate of anencephaly dropped to zero.[9]

Although we cannot provide documentation, it appears that there has been a change of attitude in a proportion of parents regarding prenatal diagnosis and its potential consequences. We wish to present data about the spontaneous outcome of pregnancy after prenatal diagnosis of anencephaly to allow obstetricians to offer better information in cases where parents are considering continuing their pregnancy. This study was prompted by the personal experience of one author (M.J.).

Methods

The first author (M.J.) was confronted with the diagnosis of anencephaly in week 22 of her fourth pregnancy. She and her husband chose not to interrupt the pregnancy and after an otherwise uneventful pregnancy she delivered a girl at term, who died 13 hours later. Based on this personal experience, M.J. created two homepages (www.anencephalie-info.org and www.prenat.ch) in autumn 2000. The aim was to provide information and support as well as an exchange of views among parents confronted with the diagnosis of an anencephalic baby. It became obvious that parents were willing to share their experience with others, including explicitly permitting publication, if data were anonymous. In this way, information was collected from 211 pregnancies. About 90% of the informants were North American (USA, Canada), the remainder were from Australia, the UK, France, Germany, Switzerland and Austria. A selection bias towards families

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with access to the internet is likely. Collection of the data was prompted by parents, and no approval of an institutional research committee was obtained. We felt that publication of this unique information in an anonymous way with the parents' permission is justified and in the interest of parents faced with a prenatal diagnosis of anencephaly. The inclusion criterion was anencephaly. We have no doubt about the correct diagnosis as prenatal ultrasound recognition is straightforward and as the parents saw their offspring after delivery. The other data considered (e.g. pregnancy duration, mode of delivery and survival) are clear cut and reliably observed by parents; confirmation by medical reports was not available and was deliberately not attempted.

The homepage (www.anencephalie-info.org) had 500 000 visits in 4.5 years, in the last few months, 28 000 visits per month, and it now provides information in English, French, German, Dutch, Portuguese and Spanish. The last author (E.B.) was contacted by M.J. as representative for NTD within the Swiss Paediatric Surveillance Unit.

Results

A summary of relevant data is given in Table 1. Polyhydramnios was noted in 56 (27%) instances, and among this group, premature delivery was more frequent (31/56) than overall (72/211). Other pregnancy complications were rare (hypertension, one; haemorrhage, one) and mothers with previous normal pregnancies noted no subjective difference in general wellbeing and fetal movements.

Prenatal death *in utero* occurred in 15 instances diagnosed from 18 to 40 weeks of gestation and in ten pregnancies at <30 weeks.

Survival of anencephalic offspring is summarised in Table 1. Of the 153 liveborn infants, 103 (67%) died within 24 hours and 41 (28%) within 1 hour. The longest survivals were 10 days (four), 18 days (one) and 28 days (one). Additional malformations, e.g. myelomeningocele, omphalocele and cleft lip and palate were present in 18 (8%) neonates. This is likely an underestimate, as it refers only to grossly visible anomalies. Duration of pregnancy (gestation) is summarised in Table 1.

Delivery by caesarean section was performed in 55 (26%) mothers. We are unable to provide clear indications in all instances; common indications were breech presentation, twin pregnancy with the anencephalic twin presenting first and placenta praevia. Many mothers asked for caesarean section with the aim of avoiding stillbirth. Six babies were delivered stillborn following caesarean section.

Spontaneous vaginal delivery did not feel different to mothers who had previously delivered a healthy baby, contrary to the belief that delivery may be prolonged due to the smaller head.

Table 1. Spontaneous outcome of 211 pregnancies after prenatal diagnosis of anencephaly (summary)

	n	%
Sex		
Female	122	58
Male	89	42
Twins (one affected)	22	10
Triplets (one affected)	1	
Polyhydramnios	56	27
Death *in utero* (prenatal death)	15	7
Gestation (weeks)		
<37	72	34
37–42	113	54
>42	21	10
Not determined	3	
Delivery, caesarean section	55	26
Survival		
Prenatal death	15	7
Stillbirth (intrapartum death)	43	20
Liveborn	153	72
Death within		
Day 1	103	49 (67% of liveborn)
In first hour	41	19 (27% of liveborn)
Second hour–24 hour	62	29 (40% of liveborn)
Day 2–5	39	18 (25% of liveborn)
Day 6–9	5	2 (3% of liveborn)
Day 10–28	6	3 (4% of liveborn)

A positive family history for NTD was reported in 12 families. Information on periconceptual folic acid substitution was not systematically recorded. The data provided are not sufficient for a sound statistical analysis, but there is an apparent trend for longer survival of an anencephalic newborn following caesarean section, while there seems to be an increased probability of stillbirth following artificial rupture of the membranes.

Discussion

It is not our intention to offer recommendations or discuss ethical and moral issues involved in management of a pregnancy with anencephaly. The motivation to compile these data resulted primarily from a lack of available information in the recent medical literature regarding the natural outcome of pregnancy in anencephaly. Although not confirmed by medical reports, we have no doubt about the diagnosis of anencephaly in these offspring, as the ultrasonographic diagnosis is straightforward and parents have seen their children. The other data reported by the parents refer to simple observations (such as duration of pregnancy, mode of delivery and survival) and can be taken as reliable.

The information gathered confirms several observations known from previous reports. There is a female preponder-

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ance among anencephalic offspring. It is also well known that polyhydramnios is more common and that anencephalic fetuses tend to deliver preterm.[10] Contrary to common belief, only a small number of anencephalic fetuses died *in utero*. More than half of the babies were born at term, 10% even after term. The high proportion of postdate gestational periods could be explained as a failure of parturition due to impaired pituitary function as previously found in anencephalic fetuses.[11] About three-quarters of anencephalic babies were born alive, but 67% of these died within 24 hours. Ten babies survived for more than 6 days. Many mothers who could compare this pregnancy and delivery with earlier experiences of normal pregnancy reported that these felt similar. Judging from these data, collected via this homepage and compared with the notion of Limb and Holmes,[9] it seems that a larger proportion of mothers carrying an anencephalic fetus are opting to continue the pregnancy rather than elective termination. From the perspective of the these mothers/parents, it is important to experience as normal a bonding as possible between mother/parent and baby and to see and touch the baby, stillborn or liveborn. It is impressive to hear from these parents who contacted our homepage that none have regretted their earlier decision to continue the pregnancy. Many parents provided photos of their children and all of the children were named. One mother opted for continuation in a subsequent pregnancy with a prenatal diagnosis of anencephaly. On the other hand, a considerable number of mothers who contacted the homepage following an elective pregnancy termination, mentioned their regret at not having seen their baby.

We hope this information is helpful for doctors and other professionals caring for parents confronted with a prenatal diagnosis of anencephaly.

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References

1 Frey L, Hauser WA. Epidemiology of neural tube defects. *Epilepsia* 2003;44(Suppl 3):4–13.
2 Botto LD, Moore CA, Khoury MJ, Erickson JD. Neural-tube defects. *N Engl J Med* 1999;341:1509–19.
3 Stevenson RE, Allen WP, Pai GS, Best R, Seaver LH, Dean J, *et al*. Decline in prevalence of neural tube defects in a high-risk region of the United States. *Pediatrics* 2000;106:677–83.
4 Yerby MS. Clinical care of pregnant women with epilepsy: neural tube defects and folic acid supplementation. *Epilepsia* 2003;44(Suppl 3): 33–40.
5 Finnell RH, Gould A, Spiegelstein O. Pathobiology and genetics of neural tube defects. *Epilepsia* 2003;44(Suppl 3):14–23.
6 Vieira AR. Birth order and neural tube defects: a reappraisal. *J Neurol Sci* 2004;217:65–72.
7 Forrester MB, Merz RD. First-year mortality rates for selected birth defects, Hawaii, 1986–1999. *Am J Med Genet* 2003;119A: 311–18.
8 Johnson SP, Sebire NJ, Snijders RJ, Tunkel S, Nicolaides KH. Ultrasound screening for anencephaly at 10-14 weeks of gestation. *Ultrasound Obstet Gynecol* 1997;9:14–16.
9 Limb CJ, Holmes LB. Anencephaly: changes in prenatal detection and birth status, 1972 through 1990. *Am J Obstet Gynecol* 1994;170; 1333–8.
10 Melnick M, Myrianthopoulos NC. Studies in neural tube defects II. Pathologic findings in a prospectively collected series of anencephalics. *Am J Med Genet* 1987;26:797–810.
11 Liggins GC, Kennedy PC, Holm LW. Failure of initiation of parturition after electrocoagulation of the pituitary of the fetal lamb. *Am J Obstet Gynecol* 1967;98:1080–6.