CASE REPORT

Prolonged unassisted survival in an infant with anencephaly

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SUMMARY

Anencephaly is one of the most lethal congenital defects. This case report is of an anencephalic infant who lived to 28 months of life and defies current literature. She is the longest surviving anencephalic infant who did not require life-sustaining interventions. This case presents the obstacles that arose from this infant's prolonged life and recommendations based on these findings.

BACKGROUND

Anencephaly is a severe neural tube closure defect that has been classified as one of the most lethal congenital defects, with a first-year mortality rate of 100%.[1] The incidence of infants born with anencephaly is 1 in 4859.[2] The Medical Task Force on Anencephaly defined this congenital defect in 1990 as having four components: an absent large portion of the skull; absent scalp over the skull defect; haemorrhagic, exposed fibrotic tissue; and absent recognisable cerebral hemispheres. Physicians screen for anencephaly during pregnancy by obtaining maternal serum α-fetoprotein levels. Ultrasounds obtained during pregnancy can also evaluate the cranial vault which reliably diagnoses anencephaly.[3] Few studies have looked at how many families choose to terminate the pregnancy, but termination rates have been reported as 43% or greater.[4 5]

Physicians counsel expectant mothers on the poor prognosis and survival rates of anencephaly based on the limited available research. When an infant survives beyond what is expected, physicians and families must decide how to best care for this critically ill infant. The following is a case of an infant born with anencephaly who contradicts the current literature and survived past 2 years of age.

CASE PRESENTATION

A gravida 2, para 1 mother aged 26 years was sent for an early dating ultrasound at 12 weeks and 2 days gestation when fetal heart tones were not auscultated by in-office Doppler. Ultrasound was suggestive of encephalocele. A first trimester screen was not performed; therefore, α-fetoprotein levels are not available. Repeat ultrasound at 14 weeks and 2 days showed the same findings. A pelvic MRI at 28 weeks gestation showed an abnormal cranio-facial ratio and microcephaly. No normal appearing brain structures were identified, including a normal ventricular system; however, there did appear to be some tissue present (figure 1). No other fetal abnormalities were identified. Maternal–fetal medicine and neonatology discussed the poor prognosis and management options with the mother. She decided to carry out the pregnancy, and donate organs and tissues after fetal demise. Routine prenatal care was provided in the family medicine centre for the remainder of the pregnancy.

A female infant was born via repeat low transverse caesarean section at 40 weeks and 2 days gestation. Birth weight was 3270 g; 1, 5 and 10 min apgar scores were 8, 9 and 9, respectively. Birth length was 48.5 cm. Physical examination revealed a flattened posterior skull and ~4 cm×4 cm occipital/parietal skull defect. Protruding from this defect was pink tissue that was neither haemorrhagic nor necrotic. She passed meconium and took formula from a bottle. She was unable to successfully breast feed but did continue to bottle feed. No heroic measures were provided to the infant to support respiratory or nutritional status, as she sustained life on her own.

After the infant survived past a few hours of life, neonatology and hospice services were consulted to discuss options, prognosis and disposition. The family elected a do-not-resuscitate code status. Routine immunisations were not given in the hospital due to anticipated short-term survival. On the third day of life, she was discharged to an inpatient hospice facility, per the family's goals of care. She continued to survive for several weeks, at which point hospice gave the family the option to take the infant home with continued palliative care. Physical examination at this time revealed a skull defect with exposed tissue protruding from that defect, not covered by scalp. She also had an

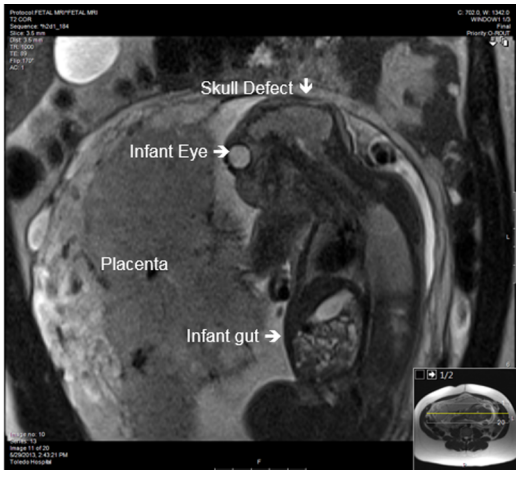

Figure 1 Twenty-eight-week pelvic MRI showing anencephalic infant.

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irregular heartbeat. The mother reported that the infant slept the majority of the day and rarely opened her eyes.

At 1 month of age, the patient was evaluated in our primary care office. She was not reaching milestones, as anticipated, but continued to breathe on her own, take formula and void and stool appropriately. At 3 months of age, she began having shaking episodes concerning for focal seizures. She was treated with phenobarbital, levetiracetam and lorazepam per hospice. The infant continued to be seen in our office every 1–2 months, and was in and out of inpatient hospice facility during the times when she appeared to be near the end of life.

OUTCOME AND FOLLOW-UP

This infant survived to 28 months of life. This photo was taken of her at about 21 months of age (figure 2). She did not require any surgical intervention nor life-sustaining treatments during her life. She lived in a home for children with developmental disabilities for the last several months of her life. Her last documented weight in our office was 9157 g which was only a 5887 g increase over her 28 months of life. She was able to feed independently, both from a bottle and at one point was tolerating pureed baby food. She also would smile spontaneously and make some cooing noises, but otherwise did not reach anticipated infant milestones. Seizures increased in frequency near the end of life, which were treated with medication prior to her passing. She was diagnosed with pneumonia and that was the eventual cause of death in combination with her other comorbidities. Her organs were unable to be donated.

DISCUSSION

Anencephaly is a lethal diagnosis, incompatible with sustained life. It has been understood that if an anencephalic infant is live-born, death will soon be imminent. Several studies have reported anencephaly to be 100% lethal in the first year of life.[1 6 7] Others reported 100% fetal demise within the first several days to weeks.[5 8 9] There have been few case reports of anencephalic infants with prolonged survival. One is a case report of two anencephalic infants who lived for 7 and 10 months, at least one of which fulfilled the Medical Task Force's diagnostic criteria.[10] Baby K was an anencephalic infant who survived 2.5 years, but unlike this case, required ventilator support.[11]

Figure 2 Photo of anencephalic infant at about 21 months of age.

This infant was diagnosed with anencephaly early in the mother's pregnancy. Family was counselled appropriately on the poor prognosis, and current documented outcomes. It was not anticipated that the infant would survive beyond the first few days of life. The family did not have clothes, diapers or means to care for this infant at home. They also had another young child and did not want to cause her undue stress. The outcome of this case should change how expectant mothers of anencephalic infants are counselled during their pregnancy. Physicians should continue to tell families of the poor prognosis; however, they also need to discuss all potential outcomes. The infant could have prolonged survival and they should be prepared to care for a critically ill infant if they choose to carry out the pregnancy.

Another component of care that was delayed was routine preventative care. Immunisations were deferred at the time of birth due to anticipated short life expectancy. After discharge from inpatient hospice, she received routine care in our family practice office. As the infant continued to survive, immunisations were offered, but the mother declined. Eventually, the infant's mother needed assistance caring for her infant. Day care facilities required her to be immunised; therefore at 12 months of age, we started a make-up immunisation schedule. The argument could be made that routine immunisations and preventive care should be provided for anencephalic infants in the case of prolonged survival. This comes down to medical decision-making between the physician and family.

When the diagnosis of anencephaly is made during pregnancy, thorough prenatal counselling should be performed. Physicians have a responsibility to tell the family of all the potential outcomes for their infant. Options for management of the pregnancy should then be given. This infant met the diagnostic criteria of the Medical Task Force on Anencephaly. Therefore, she was the longest surviving anencephalic infant who did not require any life-sustaining treatments such as intubation or feeding tubes. Knowing this rare possibility, the physician and family should make goal-oriented decisions on how to care for the infant. The provider should offer immunisations and well-

Learning points

- Physician counselling should continue to stress poor prognosis associated with anencephaly; however, all potential outcomes should be discussed, including possible, though very rare, prolonged survival.
- Parents should be counselled to be prepared to care for a critically ill infant if they choose to carry out the pregnancy.
- In the light of the possibility of prolonged survival, the physician and family should make goal-oriented decisions on how to care for the infant, including discussions of involvement of hospice care and extraordinary measures.
- In the case of prolonged survival, routine immunisations and preventive care should be provided for anencephalic infants, particularly those requiring care outside the home.

childcare to each family if the infant survives the immediate newborn period. This case should affect the practice of physicians who interact with expectant mothers of a child affected by anencephaly.

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