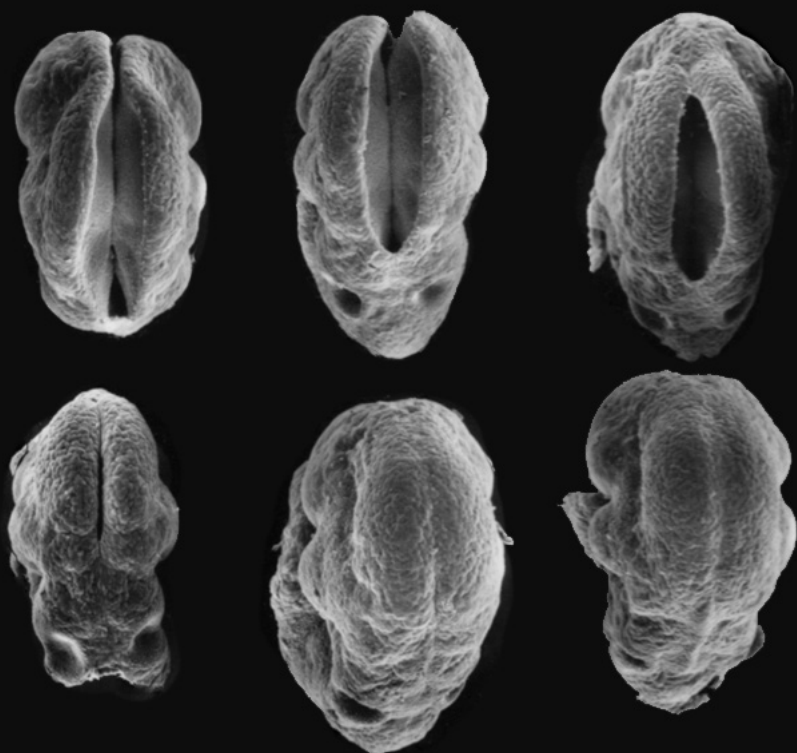


Anencephaly and  
encephalocele – two case  
reports from sub-Saharan  
Africa

June 2016



Alfonso Deliz O, Schmitt-Mechelke T, Berger TM, Rundu  
State Hospital (OA), Rundu, Namibia, Children's Hospital  
of Lucerne (SMT, BTM), Switzerland

Title figure:

source: modified from <https://w3.biosci.utexas.edu/experimentalembryology/mouseneurulation.html>

The millennium development goal (MDG) 5 aimed at improving maternal health over a 15-year time period between 1990 and 2015. MDG target 5A was to reduce the maternal mortality ratio by 75% and MDG target 5B wanted to achieve universal access to reproductive health (i.e., antenatal care, family planning, contraception) (1). In 2015, the United Nations recognized that improving maternal health was part of the unfinished agenda for the post-2015 period. The following two case reports from the Rundu State Hospital in the north of Namibia illustrate that due to the limited availability of antenatal care severe fetal malformations go unrecognized during pregnancy. Parents and health care professionals in low- and middle-income countries are therefore forced to cope with unexpected situations *ad hoc*.

## CASE REPORT 1

The mother of the first patient was a 27-year-old HIV negative G4/P4 with an unremarkable past medical history. Her first three children were alive and healthy. In the Kavango region of Namibia, routine prenatal ultrasound is not available. There was no history of polyhydramnios. The female infant was born at term by normal vaginal delivery. Findings consistent with a diagnosis of anencephaly were immediately recognized (Fig. 1, 2). The girl adapted with Apgar scores of 5, 6 and 10 at 1, 5 and 10 minutes, respectively. Her birth weight was 3600 g (P60) and her birth length was 53 cm (P85).

On admission to the neonatal unit, she was pink, had a respiratory rate of 46 breaths per minute and a heart rate of 150 beats per minute. While the baby showed movement of her extremities and was able to withdraw from painful stimuli, she had no sucking reflexes. No additional investigations were performed. The infant received intravenous fluids but no nasogastric tube feedings. The girl died on day 8 of life.



Fig. 1

*Case report 1: term infant born with anencephaly: the infant is pink and breathing spontaneously.*



**Fig. 2**

*Case report 1: term infant born with anencephaly: some ill-defined tissue protrudes through the scalp defect.*

This female infant was born by spontaneous vaginal delivery at term to a 36-year-old G4/P4 HIV negative woman with unremarkable past medical, family and social histories. During the current pregnancy, the mother had been hospitalized in a private hospital for a few days at about five months of pregnancy because of threatening miscarriage. According to the mother, an ultrasound examination obtained at the time was considered to be normal.

The baby girl adapted well with Apgar scores of 8, 10 and 10 at 1, 5 and 10 minutes, respectively. Her birth weight was 3200 g (P30). A large mass protruding from the forehead was noted (Fig. 3, 4). This lesion was not covered by skin and resembled brain tissue. The infant was admitted to the neonatal unit. On admission, she was pink, active and alert. Her respiratory rate was 50 breaths per minute and her heart rate was 145 beats per minute. She was moving all extremities and had normal reflexes; she was able to suck and swallow.

The infant was transferred to Katutura Hospital in Windhoek by ambulance (transport distance more than 700 km) for further investigations and surgical treatment. Apparently, no imaging studies were obtained and no final diagnosis was made. The baby was not operated and died at the age of 3 weeks.



**Fig. 3**

*Case report 2: term infant with a large mass protruding through the frontal region of the head.*





Fig. 4

*Case report 2: term infant with presumed encephalocele.*

## DISCUSSION

In low- and middle-income countries, many pregnant women have limited access to prenatal care; routine ultrasound screening during pregnancy is not available. The consequences are manifold and can be dramatic. Unrecognized placenta previa, for example, is associated with significant maternal and neonatal morbidity and mortality. Breech presentations in primigravid women or cephalopelvic disproportion due to fetal macrosomia can lead to obstructed labor. When Cesarean sections are not available locally and transport to a referral center is unreliable and/or very long, severe consequences for both mother and child can arise.

According to the the Millennium Development Goals (MDG) Report 2015 (1), only half of pregnant women in the developing regions receive the recommended minimum of four antenatal care visits. As pointed out above, MDG-5A aimed at reducing the maternal mortality ratio by three quarters between 1990 and 2015. In reality, maternal mortality ratio dropped by 45% worldwide during this time period (from 380 to 210 maternal deaths per 100'000 live births). Despite this progress, every day hundreds of women die during pregnancy or from childbirth-related complications. Maternal deaths are concentrated in sub-Saharan Africa and Southern Asia, which together accounted for 86% of such deaths globally in 2013. The most recent figures for Namibia (where the two patients

**Table 1**

<b>Manifestations of primary neurulation defects</b>	<b>Description</b>
<b>Craniorachischisis totalis</b>	Total failure of neurulation, no overlying axial skeleton or dermal covering
<b>Anencephaly</b>	Failure of anterior neural tube closure, frontal bones above supraciliary ridge, parietal bones and squamous part of occipital bones usually absent
<b>Myeloschisis</b>	Defect of posterior neural tube closure, involves large portions of the spinal cord and presents as a flat, raw, velvety structure with no overlying vertebrae or dermal covering
<b>Encephalocele</b>	Restricted disorder of anterior neural tube closure, occurs in the occipital region in 70 to 80% of cases (often associated with hydrocephalus)
<b>Myelomeningocele</b>	Restricted failure of posterior neural tube closure, occurs in the lumbar region in 80% of cases (almost always associated with Arnold-Chiari malformation and hydrocephalus)

*Disorders of primary neurulation (adapted from Volpe JJ) (4).*

presented in this report were born) and Switzerland are 130 and 6 maternal deaths per 100'000 live births, respectively (2), illustrating the enormous gap between developing and developed countries.

Even if mothers and infants survive labor complications, significant and sometimes dramatic long-term consequences can occur. Perinatal asphyxia is one of the leading admission diagnoses in neonatal units in many countries of sub-Saharan Africa. Without access to emergency obstetric care and skilled birth attendance, mothers may develop obstetric fistula. It has been estimated that 2 million women are living with fistula worldwide (90% in Africa). Affected women are highly stigmatized and socially isolated; access to curative surgery is frequently not available (3).

Severe fetal malformations, such as the two presented cases, are often only recognized after delivery. Both parents and health care professionals are thus ill prepared and forced to cope with such situations immediately after delivery with obvious psychological consequences.

Both patients showed disorders of primary neurulation (4). This term refers to the inductive events that occur on the dorsal aspect of the embryo and result in the formation of the brain and spinal cord during the third and fourth weeks of gestation (Table 1). The lateral margins of the neural plate invaginate

**Table 2**

<b>Clinical feature</b>	<b>Incidence in liveborn infants with anencephaly (n=12)</b>
<b>Reactive pupils</b>	25%
<b>Spontaneous eye movements</b>	33%
<b>Oculocephalic responses</b>	50%
<b>Corneal reflex</b>	50%
<b>Auditory response</b>	42%
<b>Suck, root and gag responses</b>	58%
<b>Spontaneous respiration</b>	100%

*Brainstem function in liveborn infants with anencephaly (adapted from Peabody et al) (5).*

and close dorsally to form the neural tube. The first fusion of neural folds occurs in the region of the lower medulla at approximately 22 days; closure generally proceeds rostrally (completed at 24 days) and caudally (completed at 26 days). The posterior site of closure is approximately at the lumbosacral level; the more caudal cord segments are formed by a different mechanism, a process referred to as secondary neurulation (4).

Folic acid is a synthetic form of folate used in supplements and fortified staple foods (like wheat and maize flour) to reduce the occurrence of neural tube defects (NTDs). Supplementation with folic acid is internationally recommended to women from the moment they are trying to conceive until 12 weeks of pregnancy. Another option recommended by the World Health Organization is that women of reproductive age take intermittent (weekly) iron and folic acid supplements, especially in populations where the prevalence of anemia is above 20%. Such a policy is in place in Namibia, but adherence to it is probably poor (personal communication). The most recent Cochrane Collaboration review on this topic concluded that periconceptual folate supplementation significantly reduces the risk of NTDs in comparison with no intervention, placebo or multimicronutrients without folic acid (RR 0.31, 95% CI 0.17 to 0.58) (5).

Liveborn infants with anencephaly (case report 1) frequently have some degree of brain stem function (Table 2). The prognosis for such patients, however, is dismal. In one study of 211 pregnancies, in which parents opted not to terminate pregnancy, 58 (27%) were stillborn. Of the 153 liveborn anencephalic infants, 103 (67%) died within 24 hours; only 6 survived for more than one week (maximum 28 days) (6). Because of their short life expectancy, anencephalic infants have been considered as possible organ donors for other infants. In one study, the medical care of 12 liveborn anencephalic infants was modified for one week to determine whether organ viability could be maintained and whether the criteria of total brain death could be met. Six received intensive care from birth, and six only when signs of imminent death developed. Only two infants met the criteria for total brain death within one week, and no solid organs were procured (7).

Encephaloceles (case report 2) are rare and, based on their location, can be classified as occipital (75%), sincipital (15%) or basal (10%) (8). The patient presented here most likely had a large sincipital or frontoethmoidal encephalocele; interestingly, the protruding tissue was not covered by skin. Admittedly, since no imaging studies were obtained, other diagnoses (e.g., amniotic band syndrome) cannot be excluded. Complex surgical procedures in neonates cannot be performed in Namibia; occasionally, selected patients (e.g., patients

with congenital heart disease) are transferred to South Africa for more advanced care.

Palliative care is still relatively new to national health systems, particularly in low- and middle-income countries. Significant barriers exist in the lack of clear policies establishing palliative care, lack of educational programs to teach palliative care, lack of essential medications needed to deliver palliative care, and lack of organized programs to deliver palliative care. Access to opioid medication for pain control is an enormous problem worldwide. The World Health Organization has estimated that 80% of the world's population lacks adequate access to opioid medications for pain control. Australia, Canada, New Zealand, the United States, and several European countries account for more than 90% of the global consumption of opioid analgesics (9).



1. The Millennium Development Goals Report 2015 (accessed April 13, 2016) ([\*Full text\*](#))
2. Millennium Development Goals Indicators – the official United Nations site for the MDG indicators (accessed April 13, 2016) ([\*website\*](#))
3. Fookes F - Bridgewise. Obstetric fistula: a silent tragedy ([\*website\*](#))
4. Volpe JJ. Neurology of the newborn. Fifth edition (2008), W.B. Saunders Company (no abstract available)
5. De-Regil LM, Peña-Rosas JP, Fernández-Gaxiola AC, Rayco-Solon P. Effects and safety of periconceptional oral folate supplementation for preventing birth defects. Cochrane Database Syst Rev 2015;12:CD007950 ([\*Abstract\*](#))
6. Jaquier M, Klein A, Boltshauser E. Spontaneous pregnancy outcome after prenatal diagnosis of anencephaly. BJOG 2006;113:951-952 ([\*Abstract\*](#))
7. Peabody JL, Emery JR, Ashwal S. Experience with anencephalic infants as prospective organ donors. N Engl J Med 1989;344-350 ([\*Abstract\*](#))
8. Kehila M, Ghades S, Abouda A<sup>HS</sup>, Masmoudi A, Chanoufi MB. Antenatal diagnosis of a rare neural tube defect: sincipital encephalocele. Case Rep Obstet Gynecol 2015;2015:613985 ([\*Abstract\*](#))
9. Worldwide palliative care alliance. Global atlas of palliative care at the end of life (January 2014) (accessed April 15, 2016) ([\*Full text\*](#))

SUPPORTED BY  **Vifor Pharma**

CONTACT Swiss Society of Neonatology  
[www.neonet.ch](http://www.neonet.ch)  
[webmaster@neonet.ch](mailto:webmaster@neonet.ch)