



## Anencephaly

### WHO International Classification of Diseases

Congenital malformations, deformations and chromosomal abnormalities

### OMIM Number

206500

### Mode of Inheritance

Autosomal recessive in nonsporadic cases

### Description

Incomplete fusion of the neural tube and overlying bone, soft tissue, or skin leads to several defects, varying from mild anomalies (e.g., spina bifida occulta) to severe anomalies (e.g., anencephaly). Anencephaly is a fatal autosomal recessive neural tube defect characterized by the absence of scalp, calvarium, and normal brain, which is replaced by an angiomaticous mass. Central nervous system (CNS) findings include absence of the cerebral hemispheres, the spinal cord pyramidal tracts and the cerebellum. Pathological studies reveal hypoplastic pituitaries and small adrenals, deficient in a fetal zone. The thyroid, gonads, and the number of pancreatic islets are normal, but P-cell hypertrophy is often seen. Two thirds of the anencephalic fetuses die in utero, and those that are alive at birth rarely survive more than a week. The anomaly is twice as common in females as in males.

### Epidemiology in the Arab World

#### Bahrain

Hassani et al. (2005) presented a case of inferior conjunction in a rare type of conjoined twins, dicephalus in a male fetus. The male fetus was born to a 24-year-old, gravida 2, and para 0, who had medical abortion at 15 weeks of gestation due to anencephaly with meningoencephalocele revealed by ultrasound examination. The fetus was born with 2 anencephalic heads with a bifurcation of the

vertebral column and presence of 2 spinal cords. The other viscera and limbs were normal in number and location as for a male singleton. Hassani et al. (2005) concluded that their case illustrates the relationship between conjoined twinning, and neural tube defect more particularly anencephaly with a male zygote, which is an unusual presentation for this type of zygote gender.

#### Iraq

Zlotogora (1995) stated that among families originating in Iraq, anencephaly is the most prevalent neural tube defect. He suggested the existence of a major autosomal recessive gene responsible for anencephaly in this community.

#### Kuwait

Farag et al. (1986) reported 3 sibships in 2 kindreds with multiple cases of 'nonsyndromal' anencephaly, including 2 instances of like-sex twins concordantly affected. In 1 kindred, 2 affected sibships were offspring of consanguineous parents. In 1989, Farag et al. reported a marked fall in the frequency of anencephaly among Bedouins in the last 20 years, which they attributed to a better maternal diet. A mass educational dietetic program to the Bedouin women had emphasized the importance of fresh vegetables and fruit, rich in folic acid, in addition to their traditional foods, rice and meat.

#### Morocco

Benjelloun et al. (1983) indicated a high occurrence of anencephaly in Morocco especially in males with anencephaly associated with post-maturity. Benjelloun et al. (1983) studied 13 cases of anencephaly without hydramnios in which post-maturity occurred.

#### Palestine

Dudin (1997) conducted a preliminary study to estimate the incidence of neural tube defect (NTD) among Palestinians living in East



Jerusalem and the southern part of the West Bank (600,000 inhabitants). Between 1 January 1986 and 31 December 1993, all NTD in fetuses weighing more than 500 g or of more than 22 weeks gestation, whether the product of abortion, therapeutic termination, stillborn or liveborn, were included. The study included 26,934 consecutive newborns. There were 148 cases of NTD, an incidence of 5.49 per 1000 births. The female to male ratio was 1.5:1. The incidences of spina bifida, encephalocele and anencephaly were 2.23, 0.44 and 2.41 per 1000, respectively. The incidence of NTD increased with maternal age.

### Saudi Arabia

In 1992, Rejjal and Abu-Osba reported what could be the first case of anencephaly with cleft lip and palate in a set of pergonal-induced discordant triplet pregnancy.

Al Jama (2001) reviewed 14,762 singleton live-born babies during a period of 6 years at the King Fahad Hospital, Al-Khobar, Saudi Arabia. The incidence of congenital malformations in the study was 17.0/1000 live births. Major abnormalities were present in 74.4% and minor ones in 25.6% of cases. The anatomical organs most commonly affected were the central nervous system (CNS), musculoskeletal and renal defects followed by gastrointestinal and chromosomal defects. CNS anomalies accounted for 48.8% of the total defects, hydrocephaly, anencephaly and meningocele being the predominant lesions. The incidence of malformed babies in diabetic mothers was 7.8%. Multiple anomalies were present in 16.7% infants. Of the total 251 malformed infants, 38 died within the first week of life with a mortality rate of 15.1%, compared with the overall perinatal mortality rate of 12.2/1000 live births in the hospital during the period of study.

### Tunisia

Gaigi et al. (2000) conducted an epidemiologic study for 8 years in Tunisia and calculated the incidence rate of anencephaly to be 1.15 per 1000 births (between 0.52 and 1.66 per 1000 per year). Anencephaly occurred more in female patients (sex linked = 0.59). Gaigi et al. (2000) indicated that the antenatal echographic diagnosis is efficient in depicting the disease in 90.5% of the cases.

### United Arab Emirates

Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January

1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Multifactorial disorders accounted for 26% of the cases. In their study, Al Talabani et al. (1998) observed 9 cases of anencephaly in families from the United Arab Emirates. Recurrence was not reported in other members of the families. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities in the whole United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi, the capital of United Arab Emirates.

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