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Anencephaly

Hussain Saheb S[1], Muralidhar P Shepur[2], S.D Desai[3], S.T Thomas[4], Haseena S[5]

[1,5] Research Scholars in BLDE University`s BM Patil Medical college, Bijapur, Karnataka
[3] Professor & Head of Anatomy, BM Patil Medical College, Bijapur, Karnataka.
[1,2,4.] Assistant Professors, Department of Anatomy, JJM Medical College, Davangere, Karnataka, Pin – 577004.

Abstract

Anencephaly is an embryological defect of the neural tube. The characteristic features of this are absence of cerebral hemispheres and cranial vault. In present article we have reported a case of stillborn anencephaly.

INTRODUCTION

Anencephaly is a congenital malformation in which both cerebral hemispheres are absent, there are absence of cranial vault and structures derived from forebrain and skull [1, 2]. Most of the anencephaly infants are still born, those infants born alive, die shortly after birth due to sepsis, pneumonia or atelectasis rather than from endocrine causes. In embryological aspects anencephaly results from failure of neural tube closure. It is very rare occurs 1: 20000 infants [3]. In absence of forebrain and midbrain they are replaced by fibro vascular tissue with scattered islands of neural elements. The cranial vault is defective over vertex is exposing soft angipmatous mass of neural tissue covered by a thin membrane which is continuous with skin. The cranial abnormality may extend inferiorly to the cervical region with formation of a complete spina bifida. The optic globes are usually protuberant because of inadequate bony orbits. The cerebral hemispheres, the basal ganglia and the hypothalamus are absent. If the cerebellum is present usually it is imperfect [4].

Anencephaly is a lethal condition full term infants with incomplete anencephaly who live for several days may respond to auditory, vestibular and painful stimuli. Neuroendocrine defects are frequent with failure of endocrine end organ development secondary to a hypo plastic pituitary, adrenal insufficiency may be associated with adrenocortical hypoplasia, posterior pituitary is also hypo plastic and may cause clinical diabetes insipidus. Associated abnormalities includes congenital talipus, equinovarus or valgus, spina bifida, cystic kidneys, cystic liver, high palate with cleft, diaphragmatic hernia, immature lungs, thymic hyperplasia, megaoesophagus, hypertrophy of bladder and hypoplasia of epididymis [5].

CASE REPORT

We have received a still born female fetus with anencephaly from OBG department, JJMMC, Davngere, India. We have observed in this fetus, defect of cranial vault, full absence of scalp. The brain tissue and spinal cord in the cervical region were exposed fully. The cranial and occipital region merged fully and below the cervical region it was covered by normal skin. The facial features were the eyes were very large than usual and bulged outwards, the ears were folded, nose was slightly broad, the lips were larger than usual and palate was not developed fully, other malformation were not observed(Fig 1.)

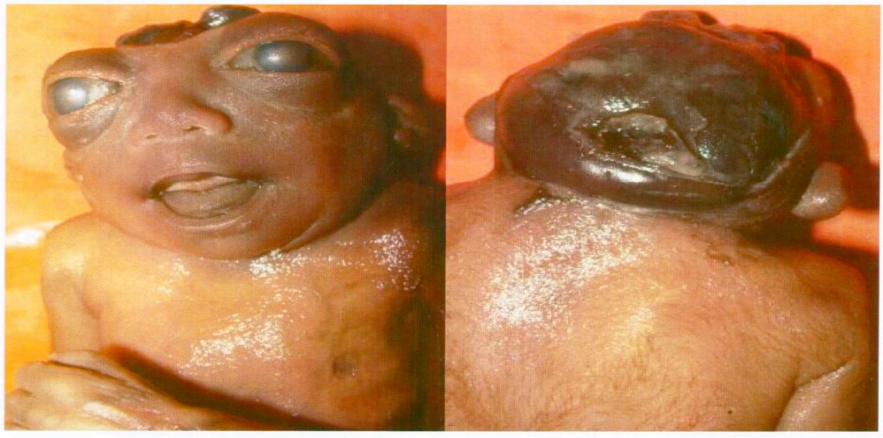

Fig 1. Showing fetus with anencephly

DISCUSSION

Anencephaly follows failure of closure of the anterior neural tube approximately at 28 days of fetal life. A less possible hypothesis proposes that anencephaly is established often a reopening of the prosenencephalic portion of the neural tube, subsequently the covering mesoderm and ectoderm incur injury. Among common neural defects the anencephaly is one of the most commonest, the incidence of anencephaly is 1:1000 to 1:20000 [3]. Epidemiology studies demonstrate variation in prevalence rates. The highest incidence is in Great Brittan and Irland and the lowest is in Asia, Africa and south America. Anencephaly occurs six times more frequent in white than in blacks, females are more often affected than males [6, 7]. Anencephaly has frequently in been found to recur in sibships. Diaphragmatic hernia is one of the associated defect with anencephaly but not usually familial [8]. Anencephaly may also associated with defect of internal organs like hypoplatic lungs, syndactyly, cyclopia, club foot, cleft palate, imperforate anus, renal defects, cardiac defects, large thymus, absence of thumb and radius, large thymus, and reduced size of adrenal gland [9].

Most of the anencephaly shows a multifactorial pattern of inheritance, with interaction of multiple genes as well as environmental factors. The specific genes are not yest been identified which can cause the neural tube defects. One such gene methylene tetraphydrofolate reductase has been shown to be associated with the rise of neural tube defects [10].

Anencephaly can be diagnosed prenatally with a high degree of certainty [11]. The initial screen for anencephaly and other neural tube defects is performed by testing for high levels of maternal serum alpha-fetoprotein in the second trimester of pregnancy and by ultrasonography in the third trimester of pregnancy [12, 13].

Anencephaly is a clearly not a sex linked malformation and its atiology is still being disputed. There is evidence however of a major gene involment in familial anencephaly with parental consanguinity and is likely to be recessive in inheritance [14, 15].

The preventive measures are very valuable and an important role seems to belong to the diet supplementation with folic acid before pregnancy and in the first month [16, 17]. This attitude has been associated with a decrease in both the frequency and in severity of the condition [18]. Another measure to be used is the fortification of both wheat and maize flour with folic acid [19]. A secondary line of prevention is to detect the abnormality as soon as possible during the pregnancy, obtained by the implementation of the program of the prenatal diagnosis [20]. The knowledge is very help for diagnose and treating of neural tube defects.

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