INIENCEPHALUS—PRENATAL DIAGNOSIS

by

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Iniencephaly is a rare foetal anomaly incompatible with life. It was first described and named by Saint-Hilaire in 1836, who reported three cases. The incidence is placed at 1.39% of all foetal anomalies by Stark (1951).

The anomaly consists of deficiency of the occiput in the region of the foramen magnum, spina bifida of considerable extent and retroflexion of the spine (Saint-Hilaire, 1836).

Lewis (1897) collected 22 cases from the world literature and classified them into three groups,

I. Iniencephalus clausus (9 cases), where there is no encephalocele.
II. Iniencephalus Apertus (8 cases) associated with small encephalocele with most of the brain in the cranial cavity.
II. Inciencephalus Apertus with a large encephalocele.

Since then, there have been sporadic case reports in the literature by Ballantyne (1904), Dorland (1925); Welz and Lieberman (1927); Bear (1937); Howkins and Lawrie (1939); Paterson (1944); and Dastur (1947). Stark, in 1951, reported two cases, bringing the total number of cases reported in the world literature to 66.

The anomaly is very rare as is borne out by the fact that out of 130,030 pregnancy terminations studied by Jayant, Mehta and Sanghvi (1960) there were only two cases of iniencephalus. Out of the 19,615 infants delivered over a ten year period at Mount Sinai Hospital there was no case of iniencephaly, though there were 296 cases of congenital abnormalities (Wacker, 1963). Ghosh and Bali (1963) found 147 cases of congenital malformations in the new-born out of 4,353 babies born at the Irwin Hospital, New Delhi, from April 1959 to July 1962, and came across no case of iniencephalus.

Hadley, Gault and Graham (1958) found 31 cases of congenital anomalies in 215 autopsy examinations done in cases of stillbirths and neonatal deaths but there was no iniencephaly in their material.

Case Report:

B., 35 years old female, 9th gavida, was referred for radiological examination for acute hydraminos when twenty-eight weeks pregnant. Her previous eight deliveries were normal; there was no history of congenital anomalies in the family.

On clinical examination—the uterus was

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the size of 36 weeks' pregnancy, foetal parts could not be made out per abdomen. Diagnosis of hydramnios was made and x-ray examination asked for to exclude any associated congenital foetal abnormality.

Fig. 1. P.A. view of the abdomen showed fundus of the uterus upto level of D12, a single small foetus was seen, presenting by the vertex but lying high up and showing very marked retroflexion of the head. A provisional diagnosis of iniencephalus was made but a repeat x-ray was asked for to confirm the diagnosis. Fig. 2. Lateral view of the abdomen taken 25 hours later showed a retroflexed head with acute angulation at the cervical spine. No soft tissue tumour could be seen on the anterior aspect of the neck to account for it. Because of the acute and persisting retroflexion of the head for which no obvious soft tissue cause was found, and suggestion of fusion of the occiput with the cervical spine and cervical spina bifida, a prenatal radiological diagnosis of iniencephalus was made.

In view of the x-ray report and the patient's discomfort due to acute hydramnios, premature induction of labour was done. A stillborn female foetus weighing 4 lbs. was delivered.

On examination—there was gross hyperextension of the cervical spine. The front of the chest was running up to the chin in a straight line erasing the normal neck folds. No other obvious abnormality was found.

Fig. 3 clinical photograph of the stillborn iniencephalic foetus.

Fig. 4 Radiograph of the foetus, anteroposterior and lateral views showing gross spina bifida of the cervical and upper dorsal spine with malformation of bodies and agenesis of the lower thoracic ribs. No encephalocele or myelocele was found.

Discussion

Iniencephaly, like other gross foetal abnormalities is incompatible with life. Like anencephaly, and hydrocephaly it is found more often in the females. Paterson (1944) found that out of the 61 cases of iniencephalus reported till then, there were 44 females, 5 males and 12 unspecified. Other associated abnormalities like cleft palate, hare-lip, talipes and umbilical hernia have been reported. In Stark's first case there was associated exomphalos, double hare-lip, cleft palate, bilateral talipes and accessory digits. No prenatal radiographs were taken. In his second case, prenatal film showed extension of the foetal head and cervical spine with breech presentation. At postmortem, in addition to iniencephalus, there was right talipes and spina bifida in the lumbar region. Similarly in the cases reported by Paterson (1944); Howkins and Lawrie (1934) there were many associated congenital abnormalities.

Potter (1957) states that iniencephalus may be found in embryos or foetuses delivered at any stage of gestation. The malformation consists of a defect of the squamous part of occipital bone which produces a greatly enlarged foramen magnum and of the absence of the laminae and spines of the cervical, dorsal and sometimes the lumbar vertebrae. The vertebrae, especially those of the cervical region, are reduced in number, irregularly fused and abnormal in shape. The spine is extremely lordotic and the neck is hyperextended so that the face looks skyward and the vertex is directed posteriorly.

In most of the reported cases diagnosis has been made postnatally after the foetus is delivered. Recently, Vatsalabai (1962) reported a case of prenatal x-ray diagnosis of iniencephalus. The patient, a 25 years old female, third gravida, was referred for x-ray examination because of the

presence of hydramnios. The first skiagram showed hyperextension of the cervical spine, a repeat skiagram 10 days later showed that there was no change in the attitude of hyperextension of the head, besides, a discontinuity in the shadow of the squamous part of occipital bone was noted with an encephalocele. A prenatal diagnosis of iniencephalic monster was made. This was confirmed postnatally. Radiograph of the foetus revealed abnormal shape of cervical vertebrae and spina bifida.

Usually the labour is uneventful, only 8 out of the 66 cases reported had difficulty in labour. This is surprising, considering the foetal attitude and almost invariable enlargement of the submento-bregmatic and occipito-frontal diameters, but the average foetal weight has been low, varying from 4 to 6 lbs. Dorland (1925) reported a case of iniencephalic foetus weighing 9 lbs. 6 ozs, causing extreme dystocia necessitating craniotomy. Prenatal diagnosis is important and the condition has to be differentiated from hyperextension of the head due to muscle spasm in a normally formed foetus. Since iniencephalus, like other congenital foetal anomalies, is often associated with hydramnios, Howkins and Lawrie (1939), and Paterson (1944) stress the importance of radiography in all cases of hydramnios to make prenatal diagnosis. A good radiograph of the maternal abdomen is essential to make prenatal diagnosis accurately and this needs extra care because of the presence of hydramnios. A repeat skiagram is usually essential to confirm the diagnosis and the presence of cervico-dorsal spina bifida, encephalocele and defect in the occiput have to be recognised on the skiagrams along with persistent retroflexion of the neck, in order to differentiate the condition from extension of the foetal head due to muscle spasm.

Summary

A case of prenatal x-ray diagnosis of iniencephalus is presented in a 35 years old multipara presenting with acute hydramnios in the 24th week of pregnancy.

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Figs. on Art Paper VII