Acta Genet Med Gemellol 34:89-93 (1985)

CASE REPORT

Received 30 January 1984
Final 10 May 1984

Twins with Acardia and Anencephaly

L. Pavone[1], K.M. Laurence[3], T. Mattina[1], G. Nuciforo[2], F. Mollica[1]
[1] *Pediatric Clinic and* [2] *Institute of Pathologic Anatomy, University of Catania, Italy, and* [3] *The Welsh National School of Medicine, Cardiff, Wales*

Abstract. The authors describe a pair of twins, one of whom showed acardia while the other had severe neural tube defect including anencephaly. This is the first observation of a fetal malformation (anencephaly) in the cotwin of an acardius and confirms the fact that even the cotwin of an acardiac fetus may be at risk of severe congenital malformations.

Key words: Acardia, Craniorachischisis, Discordant monozygotic twins

INTRODUCTION

Acardia is a very uncommon developmental abnormality occurring approximately in 1:35,000 deliveries and in less than 1% of identical twin pregnancies [16]. The anomaly seems to occur only in monozygotic (MZ) twins, because a monochorionic placenta with artery to artery and vein to vein anastomoses is necessary for the survival of the acardiac fetus [6].

Pregnancies with an acardiac fetus often result in deliveries before term, but the cotwin of the acardius is usually normal [1,14].

We recently observed a pair of twins in which one fetus showed acardia while the other had a severe neural tube defect including anencephaly. This observation confirms the fact that MZ twins may have a different outcome, and suggests that even the cotwin of an acardiac fetus may be at risk of severe congenital malformation such as neural tube defect.

CASE REPORT

The parents were healthy and unrelated, and there was no history of twinning or congenital mal-

formations in their families. The father was 29 years old, and the mother was 24 years old and primipara. During the fifth week of pregnancy she suffered from a febrile upper respiratory tract infection of short duration (2 or 3 days), but no drugs were administered.

At 36 weeks of gestation, because of premature rupture of the membranes and of breech presentation, a cesarean section was performed and a stillborn anencephalic male fetus was delivered, weighing 1,840 g and 30 cm long (Fig. 1a).

The anencephaly continued on the back with a large schisis extending to the upper lumbar portion of the rachis (Fig. 1b). On the anterior side the defect extended to the superior margin of the orbits, which were small with bulging globes. The face was grossly triangular with open lips and a protruding tongue. The neck was short and large. The limbs and face were edematous. The scrotal bursae were empty. Karyotype analysis was not performed. Permission for autopsy was refused.

A few minutes after the delivery of the anencephalus, a monochorionic, monoamniotic placenta was expelled. The placenta weighed 420 g and was infarcted (Fig. 2). Its umbilical cord contained two arteries and one vein. Attached to the placenta there was a grossly ovoid mass with a slightly bilobate appearance (Fig. 3a and b). It was attached through a narrow, short, cord-like structure departing from the central portion of the ovoid mass with several vessels largely anastomized with the umbilical cord of the cotwin through the placental surface. The mass measured 8.5 x 5 x 3.5 cm and weighed 180 g. It had a smooth, reddish bright surface. Apart from the cord, the only structure present in its ventral surface was a micropenis situated approximately 2 cm below the cord (Fig. 3a). The posterior surface was uniformly smooth (Fig. 3b). Two appendages grossly resembling limbs protruded from the two opposite poles of the ovoid mass (Fig. 3a and b). In the left one, five toes could be distinguished.

At section, two vessels (one artery and one vein) were identified in the cord. Under the skin of the ovoid mass there was a layer of subcutaneous fat of variable thickness. No organs could be recognised in the central portion of the body which only contained irregular, poorly differentiated fragments of cartilage and bone surrounded by fat, fibrous and muscular tissue.

DISCUSSION

Acardia has been said to be "the worst of all possible congenital anomalies" [2], and the word "monster" is currently employed to define it. In our opinion, this term should be definitely discarded, because of its profound psychological impact on the parents and relatives. The term "acardia" also seems inappropriate, since it focuses on only one component, though important, of the maformation complex. The abnormal fetus is sometimes called "parasitic twin", but even this definition may be psychologically disturbing. "Paratwin" would be a better definition.

Being devoid of autonomous circulation, an acardiac fetus may survive *in utero* only if it receives blood supply from a twin. The supplier twin is usually normal, while his paratwin may show various types and degrees of abnormalities, depending on which segments of the body are involved and to what extent.

Several classifications of acardiac fetuses have been proposed and are currently used, though their usefulness has recently been questioned [6]. According to Napolitani and Schreiber [9], four main types of acardia can be distinguished. In the most common type, there is a total lack of development of the cephalic portion, the fetus appearing as a trunk with recognizable lower limbs, more or less defective thorax and upper extremities, and without a head ("acardius acephalus") [5]. Less frequently, a head is also present, but only partially developed ("hemicardius" or "acardius anceps") [9]. Sometimes, on the contrary, only a head without a body develops ("acardius acormus") [9,16].

Finally, in the most severe form, the fetus appears as a "blob" or a "ball of skin" without head or limbs, and sometimes covered by hair ("acardius amorphus") [12]. This

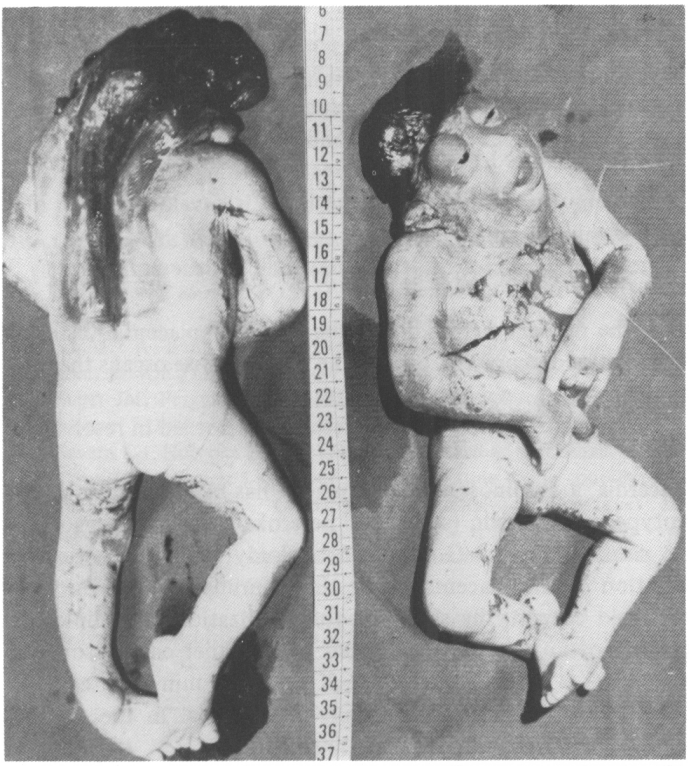
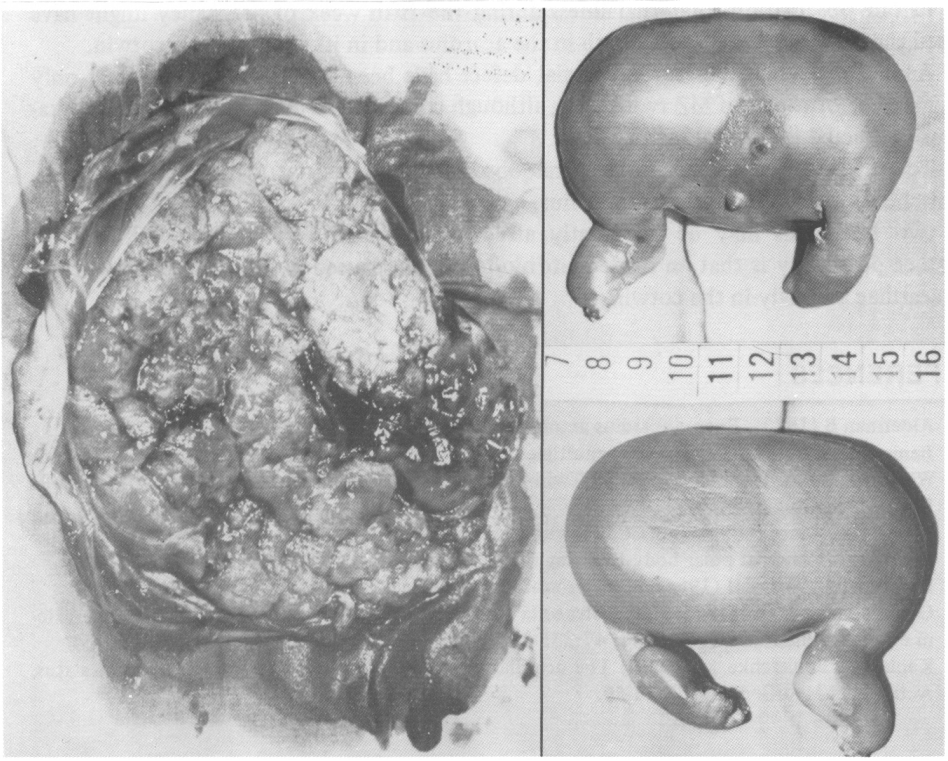


Fig. 1 - Anencephalic twin and acardius.

Fig. 2 - The placenta. Note the infarcted areas.

Fig. 3 - The paratwin.



https://doi.org/10.1017/S0001566000004967 Published online by Cambridge University Press

variety is distinguished from a dermoid cyst because of the presence of an umbilical cord and the fact that differentiated organs can be found in it [1,4,15]. The case presented here is a particularly severe form of acardius acephalus, in which the lack of development involved not only the head but also the upper extremities and most of the trunk, while the genitalia and the lower limbs were only partially developed. Looking at the pictures, one has the clear impression that only the lower half of the body is developed (even if partially), while the upper part is totally lacking.

Nothing is known about the etiological factors of acardia, and even the pathogenesis of the anomaly is uncertain.

It is still unknown whether the primary defect lies in the abnormal placental vessels (with resulting impairment of the blood supply to one of the twins, whose organs therefore fail to develop or undergo regression), or in the conceptus itself.

Cytogenetic studies performed up to now on acardiac fetuses did not aid in resolving the pathogenetic dilemma.

In at least four cases of acardia, a chromosomal abnormality has been found in the acardius, and a normal karyotype in its cotwin (so called "heterokaryotic monozygotic twinning"). Some anomalies, namely trisomy G [13] and triploidy [3,8], had clearly occurred before the differentiation of the placenta, suggesting a primary defect in the fetus. For triploid acardius-euploid cotwin, the simultaneous fertilization of a diploid first polar body and of a haploid ovum has been suggested [3,8]. Other chromosomal anomalies, such as mosaicism 46,XY/47,XY+C [11] or 46,XX/47,XX+min [15], could be either primary or secondary to insufficient blood supply and anoxia. In the other instances, the chromosome analysis has given unequivocally normal results.

In our observation, maternal illness during the fifth week of pregnancy might have caused the developmental defect both in the acardius and in its anencephalic cotwin.

Anencephaly or other severe cranial defect have been usually found to affect only one member of a pair of MZ twins [10], although conjoined twins are frequently simultaneously affected [7].

To our knowledge, this is the first observation of a fetal malformation (craniorachischisis) in the cotwin of an acardius, and suggests that the embryogenesis of each of the two MZ twins may be differently affected by a unique (environmental?) factor. Another possibility is that an inadequate blood supply caused anencephaly in a twin and the acardiac anomaly in the cotwin.

REFERENCES

1. Alderman B (1973): Foetus acardius amorphus. Postgrad Med J 49:102-105.
2. Benirschke K (1981): Lessons from multiple pregnancies in mammals. In Gedda L, Parisi P, Nance WE (eds): Twin Research 3: Part A, Twin Biology and Multiple Pregnancy. New York: Alan R. Liss, p 135-139.
3. Bieber FR, Nance WE, Morton CC, Brown JA, Redwine FO (1981): Genetic studies of an acardiac monster: evidence of polar body twinning in man. Science 213:775-777.
4. Boronov RC, West RH (1964): Monster acardius parasiticus. Am J Obst Gynecol 88:233-237.
5. Grassi G, Mitolo GR (1955): La mostruosità acefalo-acardica (contributo casistico e considerazioni etio-patogenetiche). Pathologica 47:335-359.
6. Kaplan C, Benirschke K (1979): The acardiac anomaly: New case reports and current status. Acta Genet Med Gemellol 28:51-59.

7. Moerman P, Fryns JP, Goddeeris P, Lauweryns JM, Van Assche A (1983): Aberrant twinning (diprosopus) associated with anencephaly. Clin Genet 24:252-256.
8. Nance WA (1981: Malformations unique to the twinning process. In Gedda L, Parisi P, Nace WE (eds): Twin Research 3: Part A, Twin Biology and Multiple Pregnancy. New York: Alan R. Liss, p 123-138.
9. Napolitani FD, Schreiber I(1960): The acardiac monster: A review of the world literature and presentation of 2 cases. Am J Obstet Gynecol 80:582-589.
10. Osathanondh R, Driscoll SG, Naftolin F (1975): Discordant severe cranial defects in monozygotic twins. Am J Obstet Gynecol 122:301-305.
11. Rashad MN, Kerr MG (1966): Observations on the so-called holoacardius amorphus. J Anat 100: 425.
12. Richart R, Benirschke K (1963): Holoacardius Amorphus. Report of a case with chromosome analysis. Am J Obstet Gynecol 86:329-332.
13. Scott JM, Ferguson-Smith MA (1973): Heterokaryotypic monozygotic twins and the acardiac moster. J Obstet Gynecol Br Commonw 80:52-59.
14. Severn CB, Holyoke EA (1973): Human acardiac anomalies. Am J Obstet Gynecol 116:358-365.
15. Turpin R, Bocquet L, Grasset J (1967): Etude d'une couple monozygote: fille normale-monster acardiacque féminin. Considérations anatomo-pathologiques et cytogénétiques. Ann Génét 10:107-113.
16. Warkany J (1981): Congenital malformations. In: Year Book of Medical Pathology. Chicago, p. 473-474.

Correspondence: Prof. L. Pavone, Clinica Pediatrica I, Università di Catania, Vl. A. Doria. 6, Catania, Italy.