East African Medical Journal Vol. 80 No. 11 November 2003
EAGLE-BARRETT SYNDROME: OCCURRENCE AND OUTCOMES
M. H. Aliyu, MD, MPH, Department of Epidemiology, University of Alabama at Birmingham, USA and Department of Community Medicine, Aminu Kano Teaching Hospital, Kano, Nigeria, H. M. Salihu, MD, PhD, Department of Maternal and Child Health, University of Alabama at Birmingham, USA and Department of Obstetrics and Gynaecology, Ebolowa Provincial Hospital, Cameroon and L. Kouam, MD, Department of Obstetrics and Gynaecology, University Teaching Hospital, Yaounde, Cameroon

Request for reprints to: Dr. H. M. Salihu, Department of Maternal and Child Health, University of Alabama at Birmingham, 1665 University Boulevard, Room 320, Birmingham, Alabama 35294, USA

EAGLE-BARRETT SYNDROME: OCCURRENCE AND OUTCOMES

M. H. ALIYU, H. M. SALIHU and L. KOUAM

ABSTRACT

Objective: **To examine the occurence and outcomes of African babies born with features of Eagle-Barret syndrome at a tertiary health centre.**
Design: **Case series.**
Setting: **University Teaching Hospital, Yaounde, Cameroon.**
Subjects: **Patients were identified through a retrospective review of obstetric records of mothers admitted at the centre within the period 1984 to 1996 inclusive. A total of eleven cases were identified over a period of thirteen years.**
Results: **The most prominent associated defects consisted of clubfoot, pulmonary hypoplasia, Potter's facies, imperforate anus and arthrogryposis. None of our patients survived the perinatal period.**
Conclusion: **Our study indicates the need for the establishment of a prenatal and cytogenetic infrastructure in Cameroon to enhance early detection of congenital malformation and chromosomal aberrations. In the meantime, early detection of foetuses with Eagle-Barret syndrome using ultrasound could facilitate timely institutions of antenatal management options and lead to favourable birth outcomes of affected babies.**

INTRODUCTION

The triad of Eagle-Barrett syndrome (also known as prune belly syndrome) includes absent or hypoplastic abdominal wall muscles, renal abnormalities and cryptorchidism(1). The extent to which this syndrome is encountered among African babies in the obstetric and perinatal setting is not well known. A previously published case report from Cameroon brought to our attention the importance of antenatal detection of the disorder because of the high likelihood of concomitant additional anomalies that may compromise foetal survival(2). Although ultrasound investigation is becoming popular in some major cities of Cameroon, very few obstetricians there are well-trained to diagnose congenital malformations during the early phase of foeto-genesis. As in this report, diagnosis is usually made at an advanced gestational age due to delayed referral to a tertiary centre allowing very little time for sufficient genetic counselling of affected parents. The point is therefore made for a cytogenetic infrastructure to enable adequate genetic counselling of parents and optimal management of affected patients in a developing setting like Cameroon.

MATERIALS AND METHODS

The Maternity Unit at the University Teaching Hospital (CHU) Yaounde is a referral institution receiving mostly difficult and complicated obstetric cases from peripheral hospitals and district health centres around Yaounde, the capital city. After routine examination and laboratory tests, all pregnant women seen for the first time at the centre were offered ultrasound investigation conducted either by a radiologist or a consultant obstetrician. Patients who could not pay the fees did not have it performed. In cases where a foetal anomaly was suspected, a more detailed foetal survey was done, and patients were duly informed of the diagnosis as well as their options including voluntary termination where indicated. However, a major omission in this regard is that all decisions were based on ultrasound findings without additional information from cytogenetic procedures since these could not be done. In confirmed cases of major malformations and if affected parents chose termination of the pregnancy, this was offered at the site and carried out safely. Informed consent was always sought for autopsy of the expelled foetus and placental products.

All the patients in this study were referred to and managed at CHU. The patients were identified through a retrospective review of obstetric records of mothers admitted at the centre within the period 1984 to 1996 inclusive. Patient records containing definite or presumptive diagnosis, or clinical description of a malformation were extracted from the pool and a summary of the clinical and pathological findings were prepared for each case. Three physicians were then selected: a perinatologist, a neonatologist and a pathologist. Each was asked to review the case summaries and offer a diagnosis for each patient. None of them knew what the other two were doing until all reports had been received. Where discrepancy was apparent, a solution was sought through a consensus. The patients in this study were identified from this final heterogeneous collection of congenital anomalies cases.

RESULTS

From 1984 to 1996, a total of 11 foetuses with diagnostic features compatible with Eagle-Barrett syndrome were counted. All except one case were boys (10 of 11 or 91%). Eight mothers (73%) were aged less than 30 years with most of the maternal ages clustering within the age range 20-30 years. Out of the 11 foetuses, seven (64%) were abortions occurring before the age of viability (28 weeks) of which three were induced while four were spontaneous. The remaining babies were delivered prematurely. Two of the premature babies were stillbirths while the other two survived for 3 and 48 hours respectively. Antenatal complications as well as concomitant anomalies detected by ultrasound are shown in Table 1. Ultrasound examination was conducted antenatally on seven patients, and all had abnormalities consisting of mainly oligo-anhydramnios and foetal renal system anomalies.

Table 2 is a summary of the frequency of major associated defects clinically manifested in affected foetuses. Features usually found in association with Eagle-Barrett syndrome, and which were reported among these patients included: clubfoot (five foetuses), Potter facies (four foetuses), pulmonary hypoplasia (three foetuses), imperforate anus (three foetuses), joint contractures (two foetuses), and pectus excavatum (one foetus).

Table 1

Results of maternal antenatal complications and ultrasound findings among pregnancies complicated by Eagle-Barrett Syndrome

Finding	No. (%)
Maternal antenatal complications (n=11)	
Antenatal haemorrhage	1 (9)
Hookworm anaemia	3 (27)
Preeclampsia	2 (18)
Sexually transmitted disease in pregnancy	3 (27)
Ultrasound (n=7)	
Oligohydramnios	5 (71)
Anhydramnios	2 (29)
Placenta previa	2 (29)
Renal abnormalities	7 (100)

Note: Some of the foetuses may have more than one malformation.

Table 2

Associated defects observed clinically among foetuses with Eagle-Barrett Syndrome

Associated Defect	No. %
Clubfoot	5 (45)
Potter facies	4 (36)
Pulmonary hypoplasia	3 (27)
Imperforate anus	3 (27)
Arthrogryposis	2 (18)
Pectus excavatum	1 (9)
Hypoplastic lower limbs	1 (9)
Bilobar right lung	1 (9)

Note: Some of the foetuses may have more than one malformation

Table 3

Frequency of main autopsy findings in foetuses with Eagle-Barrett Syndrome (n=9)

Autopsy Finding	No. (%)
Anasarca/ascites	2 (22)
Atrophic abdominal muscles/ midline defect	9 (100)
Cryptorchidism	6 (66)
Omphalocele	2 (22)
*Renal abnormalities	9 (100)
Single umbilical artery	2 (22)
Tracheoesophageal fistula	1 (11)
Undescended testis	4 (44)
Ventricular septal defect	2 (22)

*The most common renal abnormalities include hydronephrosis, hydroureters, renal aplasia, polycystic kidneys, and megalocystis. Some foetuses may have more than one malformation.

However, more details became available following post-mortem examination, which was accepted and carried out on nine foetuses (Table 3). Combining the information gathered from clinical, ultrasound and post-mortem examinations of these foetuses, certain categorisation was feasible. Three foetuses presented with features that could be classified as syndromes. One of the foetuses revealed two (polycystic kidney and hexadactyly), of the three criteria for Meckel's syndrome and instead of encephalocele, had microcephaly. Although clinical features of a trisomic syndrome (including Simian crease) were apparent in another foetus, this could not be confirmed due to absence of cytogenetic facilities. The sole female patient in our series was affected by severe extra anomalies consistent with both Eagle-Barrett syndrome and the VACTERL-sequence.

DISCUSSION

Eagle-Barrett syndrome (or Prune Belly syndrome) is a rare disorder occurring with a frequency of one in every 50,000 live births(3). It is characterised by the triad of cryptorchidism, urinary tract abnormalities, and absent or hypoplastic abdominal wall musculature(1). Osler is credited with being the first to describe the clinical features of prune belly in 1901(4) while Eagle and Barrett subsequently defined it as a clinical syndrome in 1950(5). The preponderance of boys among our cases (91%) is consistent with the syndrome, which is known to manifest primarily in boys(5-7). Eagle-Barrett syndrome in females tends to be associated with additional severe birth defects(8,9), as observed in our sole female foetus. It is also interesting to note that most of the mothers were younger than 30 years (73%), thus, supporting other reports linking young maternal age and prune belly(9,10). However, this observation in our study could also have been coincidental since African women generally become pregnant early in their reproductive life history.

Ultrasound investigation was performed antenatally in seven of the eleven foetuses, and all reported

abnormal results. The commonest ultrasound feature was oligoanhydramnios, which was observed in all the patients (five cases of oligohydramnios and two cases of anhydramnios). This is the most frequent ultrasound sign of PBS(11), and in our opinion, the easiest to detect even by the least experienced sonologist. The appearance of oligoanhydramnios should therefore, kindle a suspicion for possible prune belly syndrome, requiring detailed foetal survey to exclude additional anomalies.

Chromosomal aberrations have been reported in association with prune belly, and include some relatively well-known aneuploidies, such as Turner's syndrome (12-15,18,21). We found one case of clinical suspicion of a trisomic syndrome but could not confirm it by chromosomal analysis. This illustrates the necessity for the establishment of a cytogenetic infrastructure in Cameroon without which optimal genetic counselling and patient management may not be attainable.

It is apparent from our results that induced abortion was performed late in the second trimester in mothers who opted for pregnancy termination following the detection of foetal malformations (average gestational age = 24 weeks). From the case notes, this could be attributed to late referrals. Although our centre was well equipped to manage these cases, it is preferable for therapeutic abortions to be performed well before this time to minimise the risks for complications. Earlier diagnosis using ultrasound also allows for sufficient time to conduct genetic counselling.

This study appears to suggest high perinatal mortality associated with Eagle-Barrett syndrome at the study site. It is doubtful whether this is in reality the case because milder variants probably escaped detection perinatally. Furthermore, being a referral station, our centre most likely received cases of congenital birth defects that were complicated bearing a lower than normal likelihood for survival.

In summary, we report one of the few case series of Eagle-Barrett syndrome from a developing country. The most prominent findings among our patients consisted of clubfoot, pulmonary hypoplasia, Potter's facies, imperforate anus and arthrogryposis. Early detection of foetuses with prune belly using ultrasound in developing countries could potentially facilitate timely institution of antenatal management options that would lead to favourable birth outcomes of affected babies.

ACKNOWLEDGEMENTS

To the nursing staff at the Younde University Teaching Hospital and the Ebolowa Provincial Hospital for their assistance during the data collection. We are also greateful to the physicians that participated in the ascertainment of the diagnosis.

REFERENCES

1. Parker, R.W. Absence of abdominal muscles in an infant. *Lancet.* 1895; **1:**1252.
2. Salihu, H.M., Tchuinguem, G. and Ratard, R. Prune belly syndrome coexisting with the VACTERL-complex in the background of antenatal ingestion of traditional concoction. *J. Obstet. Gynaecol.* 2000; **20:**95-96.
3. Garlinger, P. and Ott, J. Prune belly syndrome. Possible genetic implications. *Birth defects.* 1974; **10:**173-180.
4. Osler, W. Congenital absence of the abdominal musculature with distended and hypertrophied urinary bladder. *Bull Johns Hopkins Hosp.* 1901; **12:**331-333
5. Eagle, J.F. and Barrett, G.S. Congenital deficiency of abdominal musculature with associated genitourinary abnormalities: a syndrome. Report of nine cases. *Pediatrics.* 1950; **6:**721-736.
6. Rogers, L.W. and Ostrow, P.T. The prune belly syndrome. Report of 20 cases and description of a lethal variant. *J. Pediatr.* 1973; **83:**786-793.
7. Sanni, R.B., Coulibaly-Denoulet, D., Coulibaly, B., Nandiolo, R., and Mobiot, M.L. The treatment of prune belly syndrome at the University Hospital Center at Abidjan (Treichville). *Ann.Urol.* 1998; **32:**23-27.
8. Lubinsky, M.S. Female pseudohermaphroditism and associated anomalies. *Am. J. Med Genet.* 1980; **6:**123-126.
9. Druschel, C.M. A descriptive study of prune belly in New York State, 1983 to 1989. *Arch. Pediatr. Adolesc. Med.* 1995; **149:**70-76.
10. Baird, P.A. and MacDonald, E.C. An epidemiologic study of congenital malformations of the anterior abdominal wall in more than half a million consecutive live births. *Am. J. Hum. Genet.* 1981; **33:**470-478.
11. Gnirs, J., Boos, R., Waldherr, R. and Schmidt, W. Das Prune-Belly-Syndrom: Pranatale Diagnose und geburtshilfliches Vorgehen. *Z Geburtsh u Perinat* 1988; **192:**114-121.
12. Lubinsky, M., Doyle, K. and Trunca, C. The association of a prune belly syndrome with Turner's syndrome. *Amer. J. Dis. Child.* 1980; **134:**1171-1172.
13. McKeown, C.M.E. and Donnai, D. Prune belly in trisomy 13. *Prenat Diagn.* 1986; **6:**379-381.
14. Frydman, M., Magenis, R.E., Mohandas, T.K. and Kaback, M.M. Chromosome abnormalities in infants with prune belly anomaly: association with trisomy 18. *Amer. J. Med. Genet.* 1983; **15:**145-148.
15. Amacker, E.A., Grass, F.S, Hickey, D.E., and Hisley, J.C. Brief clinical report: an association of prune belly anomaly with trisomy 21. *Amer. J. Med. Genet.* 1986; **23:**919-923.