Case Report

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Paediatrics Section

Eagle-Barrett Syndrome: A Case Report

CHAPAY SOREN, PARAMANANDA PUJHARI, P.V. SUBBARAO

ABSTRACT

Eagle-Barrett syndrome is a rare congenital anomaly of uncertain aetiology almost exclusive to males. It is characterized by the triad of absent or incomplete abdominal musculature, undescended testes, and urinary tract abnormalities. A male baby with above characteristics triad was born in our hospital. A diagnosis of Eagle-Barrett Syndrome was made. This was undertaken in order to highlight the occurrence of this rare syndrome in our environment and to review its pathogenesis, presentation and management approach.

Key Words: Urinary, Paediatrics, Renal Failure

INTRODUCTION

Eagle-Barrett Syndrome, also known as triad syndrome or Prune belly syndrome, represents a spectrum of anomalies with variable degrees of severity predominantly affecting males. It is a rare condition characterized by the triad of absent or incomplete abdominal musculature, bilateral cryptorchidism and urinary tract abnormalities. It is caused by urethral obstruction early in development resulting in massive bladder distension and urinary ascites, leading to degeneration of the abdominal wall musculature and failure of testicular descent. The impaired elimination of urine from the bladder leads to oligohydramnios, pulmonary hypoplasia, and Potter's facies. The urinary tract may have variable degrees of hydronephrosis, renal dysplasia, dilated tortuous ureters, an enlarged bladder, and a dilated prostatic urethra. The exact aetiology of EBS is unknown.The case is reported for its rare congenital abnormality.

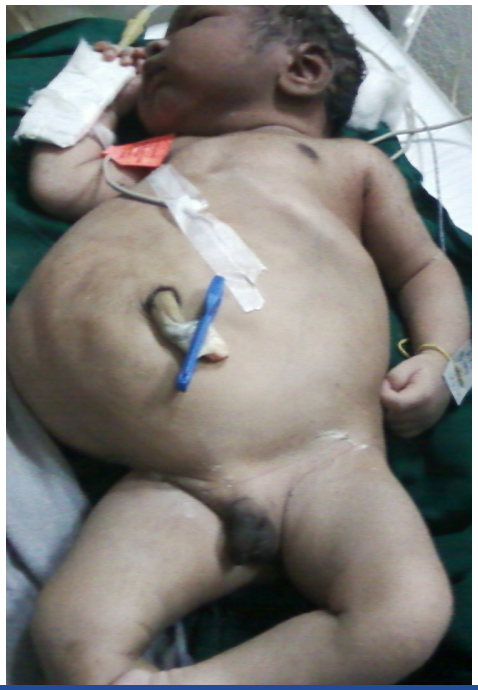
CASE REPORT

A 3.5kg male baby was delivered in our hospital by an un-booked 29-year old Gravida 2, Para 1, Live 1 mother with an APGAR score of 5 at 1minutes and 10 at 5 minutes after birth. Abdominal examination revealed huge distension with thin and wrinkled skin protruding most prominently in the right side with visible bowel loops.(figure1) Both the kidneys are palpable. Perineal examination showed bilateral cryptorchidism and hypospadiasis. Cardiac examination was normal by clinical examination and echocardiography. Investigations showed hyponatremia with Na of 130meq/l, urea of 65mg/dl, and creatinine of 1.5mg/dl. Ultrasound imaging showed mild hydronephrosis with gross dilatation pelvicalyceal system of right kidney and moderate hydronephrosis of left kidney with grossly dilated ureters, and distended bowel loops.

DISCUSSION

Eagle-Barrett syndrome is a rare congenital disorder predominantly affecting males with male:female ratio being 2.0:1 [1]. The incidence has been reported to be ranged 1 in 29,000 to 1 in 40,000 live births [2]. Parker *et al*. first recognized the three components of this syndrome [3]. The characteristic triad consists of absent or incomplete abdominal musculature, bilateral cryptorchidism and urinary tract abnormalities [4, 5]. The exact aetiology is not known, however some of the studies reveal the possibility of genetic inheritance [6]. Along with the classical triad broad spectrum of defects including musculoskeletal, cardiovascular, pulmonary, gastrointestinal and genital malformations have been documented [7]. When the urinary tract maldevelopment is associated with severe obstructive uropathy, this syndrome can lead to oligohydramnios and pulmonary hypoplasia.

The pathogenesis of Eagle-Barrett syndrome is not clearly known. The mesodermal defect theory suggests that a defect exists in the mesoderm of the anterior abdominal wall and urinary tract. Between 6 and 10 weeks of gestation, aberrant development of the derivatives of the first lumbar myotome leads to a patchy muscular deficiency or hypoplasia of the abdominal wall as well as to urinary tract abnormalities [8, 9]. An alternate theory, the urethral



[Table/Fig-1]: Baby with Eagle-Barrett Syndrome showing protruded abdomen with thin and wrinkled abdominal skin and visible bowel loops

obstruction malformation complex, proposes that pressure atrophy of the abdominal wall muscles occurs when urethral obstruction leads to massive distension of the bladder and ureters. Bladder distension would also interfere with descent of the testes and thus be responsible for the bilateral cryptorchidism. This mechanism is responsible for the urinary tract dilatation and distension [10]. The higher incidence of this syndrome in males has been explained on the basis of the more complex morphogenesis of the male urethra, possibly resulting in obstructive anomalies at several levels. Ultrasound, plain X-ray, and intravenous pyelogram are more useful investigations to diagnose the condition.

Many neonates with Eagle-Barrett syndrome have difficulty with effective bladder emptying because the bladder musculature is poorly developed, and the urethra may be narrowed. When no obstruction is present, the goal of treatment is the prevention of urinary tract infection with antibiotic prophylaxis. When obstruction of the ureters or urethra is demonstrated, temporary drainage procedures, such as a vesicostomy, may help to preserve renal function until the child is old enough for surgery. Urinary tract infections occur often and should be treated promptly. Correction of the undescended testes by orchidopexy can be difficult in these children because the testes are located high in the abdomen and is best accomplished in the first 6months of life. Reconstruction of the abdominal wall (abdominoplasty) offers cosmetic and functional benefits.

The prognosis ultimately depends on the degree of pulmonary hypoplasia and renal dysplasia. The most common complication is chronic renal failure that occurs in 25–30% of cases. Many infants are either stillborn or die within the first few weeks of life from severe lung or kidney problems, or a combination of congenital anomalies. There are cases of Eagle-Barrett syndrome who survived into adult life after abdominal reconstruction and urinary tract repair [11].

There is no known prevention but the routine use of screening for fetal anomalies is helpful. If an antenatal diagnosis of urinary obstruction is made it may be possible to perform intra-uterine surgery to prevent the development of Eagle-Barrett syndrome [12]. Early diagnosis of this syndrome and determining its optimal treatment are very important in helping to avoid its fatal course. These patients need multidisciplinary management of a neonatologist, nephrologists, and pediatric urologist for an optimal outcome.

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AUTHOR(S):

1. Dr. Chapay Soren
2. Dr. Paramananda Pujhari
3. Dr. P V Subbarao

PARTICULARS OF CONTRIBUTORS:

1. Assistant Professor of Pediatrics,
2. Assistant professor of Pediatrics,
3. Professor & Head of Pediatrics,
   Konaseema Institute of Medical Sciences, Chaitanya Nagar Amalapuram, East Godavari dist, Andhra Pradesh-533201, India.

NAME, ADDRESS, TELEPHONE, E-MAIL ID OF THE CORRESPONDING AUTHOR:

DR. Chapay Soren MD, Assistant Professor of Pediatrics Konaseema Institute of Medical Sciences, Chaitanya Nagar Amalapuram, East Godavari dist, Andhra Pradesh-533201, India.
Phone: 09440106344 ,E-mail: drcsoren55@yahoo.co.in

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