Volume : 3 | Issue : 4 | Apr 2014 | ₹ 250/-

PEER REVIEWED & REFFERED INTERNATIONAL JOURNAL
ISSN - 2250-1991 | IMPACT FACTOR - 1.6714

PARIPEX - INDIAN JOURNAL OF RESEARCH

Listed & Indexed in
International ISSN Directory, Paris

Journal for All Subjects

Volume : 3 | Issue : 4 | April 2014 ISSN - 2250-1991

Research Paper Medical Science

Rudimentary Thymus In A Case Of Potter's Syndrome

Dr. Prasenjit Bose	Department Of Anatomy, Institute Of Medical Sciences, Banaras Hindu University, Varanasi, Uttar Pradesh, 221005, India.
Dr Royana Singh	Associate Professor, Department Of Anatomy, Institute Of Medical Sciences, Banaras Hindu University, Varanasi, Uttar Pradesh , 221005, India.
Dr. Anjali Rani	Assistant Professor , Department of Obstetrics and Gynaecology, Hindu University, Varanasi, Uttar Pradesh , 221005, India

ABSTRACT

Potter syndrome refers to a group of characteristic facial findings associated with lack of amniotic fluid and kidney failure in an unborn infant. It is related to a chain of events that may have different beginnings (absent kidneys,cystic kidneys,obstructed ureters),but which all end with the same conclusion (oligohydramnios).Rudimentary thymus is a rare finding in Potter syndrome.We report here a case of 20 weeks old male fetus with Potter syndrome, who had polycystic kidneys, hypoplastic lungs, hepatomegaly, intestinal obstruction and most interesting finding was rudimentary thymus which is a rare finding.

KEYWORDS Potter syndrome, Polycystic kidneys, Hepatomegaly, Rudimentary thymus, Pulmonary Hypoplasia.

Introduction

In the year 1946, Edith Potter described a series of 20 cases with absent kidneys, noting the characteristic appearance of lungs and head (Potter,1946). She found out that all these cases have one common finding that is oligohydramnios and it was due to absent kidneys. Potter symdrome is a rare disorder with an incidence of 1 in 4,000 births with predominance in males (Potter, 1946) (Thomas, 1974). It is accompanied by severe oligohydramnios, renal abnormalities (bilateral renal agenesis, severe hypoplasia, dysplasia, polycystic kidney, and obstructive uropathy), or chronic leakage of amniotic fluid (oligohydramnios sequence) during middle gestational weeks. Renal failure is the main defect in Potter's syndrome. Later it was proved that the pressure effect of growing uterus without the presence of the amniotic fluid upon the fetus resulted in the typical facial appearances and the position of the limbs. The presence of the typical facial appearance is also known as Potter phenotype (Thomas , 1974) (Robertson and Rennie, 1999) (Kleinman, 1992).

Potter's syndrome occurs in sporadic and autosomal recessive forms (Robertson and Rennie 1999) (Kleinman 1992). The Potter syndrome can be classified into five types-1. Classic Form: In this the infant has bilateral renal agenesis. This is the most common type.2. Type I: It is due to Autosomal Recessive Polycystic Kidney Disease. Mainly associated with fibrosis or cystic changes of liver and pancreas.3. Type II: In this one kidney is absent and the other is small and malformed.4. Type III: It is due to Autosomal Dominant Polycystic Kidney Disease. Linked to mutation in genes PKD1 and PKD2. Mainly associated with hepatic cysts and enlarged spleen.5. Type IV: This occurs when a longstanding obstruction in either the kidney or ureter leads to hydronephrosis.

Other characteristic features include premature birth, breech presentation, a typical facial appearance (Potter's facies), and limb malformations. Most infants are still born or if live born die due to severe respiratory insufficiency. Clinical and Ultrasonography findings along with a positive family history are diagnostic features of the Potter's syndrome (Bain et al ,1964) (Scott and Goodburn, 1995).

Though Potter's syndrome is rare but we suggest that it may be more common because infants are stillborn , die soon after birth or most of the women in this region still do no go for hospital delivery.

Case Report

A female patient of Indian origin on routine examination for amenorrhea of 20 weeks visited the Department of Obstetrics and Gynaecology. She had undergone abdominal sonography. USG report confirmed polycystic kidneys in the developing fetus, thus mother was advised termination of pregnancy. Termination was done under aseptic procedures.

After taking proper consent from parents, the fetus was procured by the Department Of Anatomy. On external features examination, the foetus was a male with well-developed penis and scrotal sac.The typical facial appearance of potter phenotype was observed. It had widely separated eyes, broad nasal bridge,flattened nose low set ears and micrognathia (Fig 1.Arrow (white)).The following findings were observed after autopsy:thorax; hypoplasia of both lungs, well-developed heart and arterial branches (Fig. 1 black arrow) and most interestingly the rudimentary thymus. Abdominal cavity exhibited hepatomegaly with embedded gallbladder (Fig. 1.black arrow), intestinal obstruction showing dilated loop of the duodenum, meckel's diverticulum the differentiation of the gastrointestinal tract with colon atresia . Both the kidneys were polycystic , the left being small than that of right kidney. The ureters of both the side descended down to open into the posterior aspect of the urinary bladder. The lower abdominal cavity had two well-developed testis (Fig. 1).