International Journal of General Medicine
and Pharmacy (IJGMP)
ISSN(P): 2319-3999; ISSN(E): 2319-4006
Vol. 3, Issue 5, Sep 2014, 25-28

International Academy of Science,
Engineering and Technology
Connecting Researchers; Nurturing Innovations

"A CASE REPORT OF POTTER SYNDROME"

SUNANDA BAI G & DEEPASHREE G

Department of Obstetrics and Gynecology, Bengaluru Medical College & Research Institute, Bangalore, Karnataka, India

ABSTRACT

The Potter syndrome has been estimated that 1 in 40 or 2.5 percent of new borns have recognizable malformation (fifty percent single malformation & other fifty percent multiple malformations).In 2001, birth defects accounted for 1 in 5 infant deaths in United States, which is higher than other causes such as Preterm labour/ low birth weight/sudden infant death syndrome/maternal complication of pregnancy and respriratory distress syndrome [(8)]. The birth malformations may be the following reasons-Malformation syndrome due to recognizable genetic causes i.e.trisomy, Teratogenes, dysplasia sequence. i.e., Neurocutenous melonosis, Dysruption sequence i.e., Amniotic membrane rupture, Deforamtion sequence i.e., Uterine forces combined with oligohydromnios causing deformation of limbs, face, chest, etc. and malformation sequence i.e., DiGeorge sequence. Human Malformation and dysplasias are caused by the combination effects of genes and environmental factors. About 33% malformations are genetic and about 6 – 7% of malformations are due to known environmental factors such as maternal diseases, infections and teratogenic genes. About 60-70% of malformations are of unknown etiology [(8)]. A comprehensive and rational diagnostic approach can be derived from etiological principles. Hence a rare case of "POTTER SYNDROME" is discussed here.

KEYWORDS: Pregnancy, Potter Syndrome, Genetics, Oligohydromios

INTRODUCTION

Potter syndrome refers to the typical physical appearance and associated pulmonary hypoplasia of a neonate as a direct result of oligohydramnios and compression while in utero. The term was coined after the pathologist Edith Potter, who in 1946 described the facial characteristics of infants with bilateral renal agenesis [(1)]. From her research, she was able to deduce the sequence of events that led to these features. Other conditions resulting in oligohydramnios, such as obstructive uropathy, cystic kidney diseases, renal hypoplasia and primitive rupture of membranes lead to the same clinical findings. Amniotic fluid deceases as a result of leakage or inadequate production due to poor fetal urine output. Hence, the terms Potters sequence or oligohydramnios sequence emerged. Regardless of the root cause for oligohydramnios, the term Potter Syndrome, Potter Sequence and Oligohydramnios Sequence are used interchangeably in the published literature.

Renal failure is the main defect in Potters Sequence. Other Characteristic features include premature birth, breech presentation, and atypical facial appearance and limb malformations. Severe respiratory insufficiency leads to a fatal out come in most infants [(1)]. Here is a case report of a Potters Sequence.

Aims of the Study

Understanding the etiology of birth malformation is critical in evaluation and management of birth defects. Potter Syndrome is a rare case.

Material and Methods

A referred case of Potter syndrome admitted in the Department of Obstetrics and Gynecology at Bowring and Lady Curzon Hospital, Bengaluru Medical College & Research Institute is taken.

Case Report

A twenty six years old woman, second gravida, of 26 weeks gestational age, presented with decreased fetal movements and ultrasound report showing an anomalous baby. Delivered a still born female baby by breech presentation. The baby weighed 1.2kg. Placenta and membranes expelled with no liquor. Ultrasound examination at 21 weeks and 2 days showed enlarged and hyper echoic kidneys suggestive of polycystic kidney disease, empty bladder, narrow thorax, distended abdomen as a result of large kidneys and anhydramnios(AFI-O). Figure (1): Phenotypic appearance of pottery syndrome

Figure 1: Phenotypic Appearance of Pottery Syndrome

Typical Potters facies was noted which included flattened nose, recessed chin (FIGURE), prominent bilateral epicanthal folds (FIGURE) and low set ears with wide pinna. There was also presence of short lower limbs and congenital talipes equino varus(CTEV) (FIGURE)

DISCUSSIONS

Oligohydramnios typically diagnosed during the 2nd or 3rd trimester of the pregnancy. When the oligohydramnios is severe enough and is present for an extended period of time.Approximately 50% of the time, fetal renal system abnormalities causes severe oligohydramnios, resulting in the fetus developing oligohydramnios sequence. Decrease in the volume of amniotic fluid may be due to decreased urine production secondary to bilateral renal agenesis, obstruction to urinary tract or occasionally prolonged rupture of membranes. The resulting oligohydramnios is the cause of deformities in Potter sequence [(2)]. The ultimate result is decreased amniotic fluid which results in typical features that we have noticed in our case.A retrospective analysis of children with Potter Syndrome found that 21% had bilateral renal agenesis, 47% had cystic dysplasia, 25% had obstructive uropathy and 5% had other defects. Posterior urethral valves were the most common cause of bladder outlet obstruction (60%)[(3)] .Potter syndrome is mostly associated with obstruction of the urinary tract or sever bilateral renal hypoplasia. Bilateral renal agenesis is estimated to occur in about 1 of 5000 fetuses [(4)] and is responsible for 20% of Potter cases. The frequency of other causes of Potter Syndrome is not known. The associated maternal high risk factors for bilateral renal agenesis or maternal body mass index greater than 30, smoking and binge drinking[(5)(6)]. The evaluation of patients with the Potter Sequence should include an examination for non renal defects, autopsy, chromosome analysis and renal ultra sound or urologic evaluation of parents. Ultrasonographic prenatal monitoring of subsequent pregnancies is such families is strongly warranted because of a definite but unknown degree of recurrence risk [(7)].

This case is published to highlight the rarity of Potter Sequence. Regular follow up and antenatal check-up should be done to diagnose oligohydramnios to prevent the complications resulting from it. Not all oligohydramnios cases will lead to Potter Sequence, but careful examination of the fetus and regular amniotic fluid index should be done to prevent the consequence. However we lost this patient for further counselling.

CONCLUSIONS

Intervened rare case – report is helps to researcher and clinician to rule out the new cases for both rural and urban set up. More research could be needed to focus the issue of potter syndrome.

ACKNOWLEDGEMENTS

We acknowledge the help extended by Dr.Umadevi, Professor and HOD, Department of OBG, BMCRI, Bengaluru.

Conflict of interest: No financial help received from any quarters in preparing this manuscript.

REFERENCES

1. Khatami F. Potter syndrome: A study of 15 patients. Arch Iranian Med 2004;7:186-9
2. Dhundiraj KM. Madhukar DN, Ambadasrao PG, Wamanrao KS, Prem ZM. Potter syndrome: A report of 5 cases. Indian J Pathology Microbiology. 2006;49:254-7
3. Chow JS, Benson CB, Lebowitz RL. The Clinical significance of an empty renal fossa on prenatal sonography, J Ultrasound Med. Aug 2005;24(8):1049-54; quiz 1005-7
4. Slikers JE, Olshana AF, Siega-Riz AM, Honein MA, Aylsworth AS. Maternal Body mass index and lifestyle exposures and the risk of bilateral renal agenesis or hypoplasia: the National Birth Defects Prevention Study. Am J epidemiology. Dec 1 2008;168(11): 1259-67
5. Klaasen I, Neuhaus TJ, Mueller-wiefel DE Kemper MJ. Antenatal oligohydramnios of renal origin: long term outcome. Nephrology dial transplant. Feb 2007; 22(2):432-9.
6. Paces-Fessy M, Fabre M, Lesaulnier C, Cereghini S. Hnf1b and Pax2 Cooperate to control different pathways in kidney and ureter morphogenesis. Hum Mol Genet. Apr/2012;24.
7. Curry CJ, Jensen K, Holland J, Miller L, Hall BD. The Potter Sequence: a Clinical analysis of 80 cases. Am J Med Genet. 1984; 19:679-702.
8. Nelson Textbook of Pediatrics, 18th Edition.