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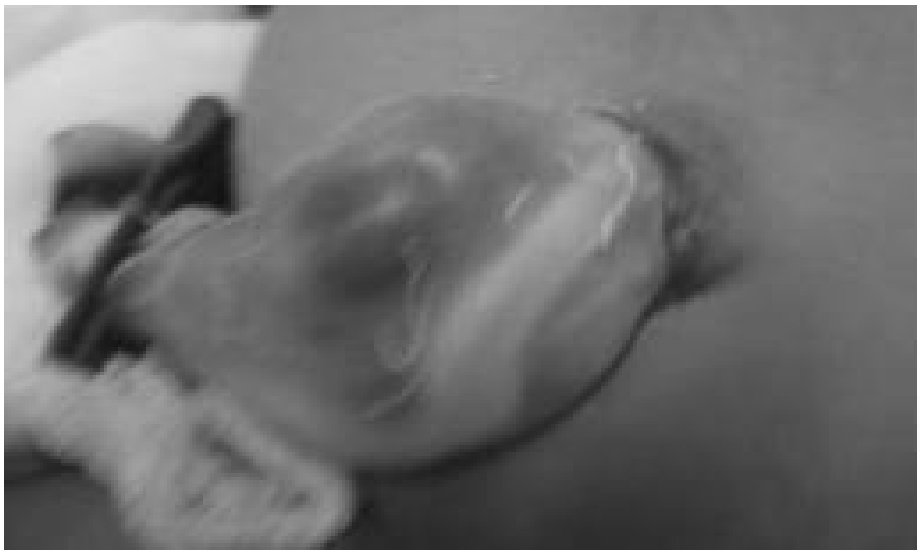

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CASE REPORT

BECKWITH – WIEDEMANN SYNDROME: AN AUTOPSY REPORT

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INTRODUCTION

Beckwith Wiedemann Syndrome is a rare entity characterised by a varied combination of congenital malformations like exomphalos, macroglossia, gigantism (visceral and somatic), cryptorchidism, craniofacial dysmorphism, anomalies of ear and neonatal hypoglycemia. We present a rare case of Beckwith - Wiedemann Syndrome in a 21 day old preterm male baby.

Case History

A 21 day preterm male baby was referred for autopsy after surgical correction of omphalocele. The baby was conceived after assisted reproduction technology and was delivered at 36 weeks of pregnancy with an omphalocele (Fig. 1). The patient had repeated episodes of hypoglycemia.

Autopsy findings: External examination revealed macroglossia. Bilateral naevus flammeus over eyelid (Fig.2),cryptorchidism seen. The scar of operated omphalocele was seen which was healthy. In situ and systemic examination revealed visceral organomegaly (hepatomegaly, enlarged kidneys), cystic right adrenal gland (Fig.3). Microscopic sections of the kidney reveal dysorganisation of parenchyma glomerular neogenesis and increased number of immature collecting tubules (Fig.4). Pancreas shows increased number of acini (Fig.5). Adrenals showed cytomegaly (Fig.6) Lungs revealed haemorrhagic bronchopneumonia (Fig.7) which was the cause of death.

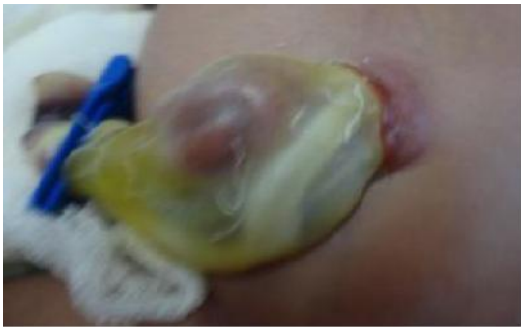

Fig. 1 Omphalocele (pre-operative)

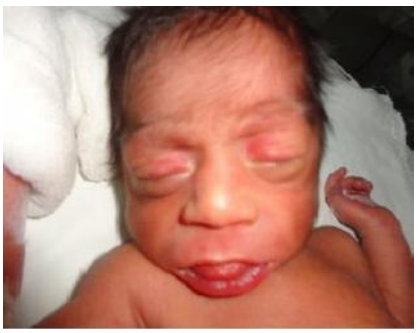

Fig.2 Bilateral naevus flammeus over eyelid

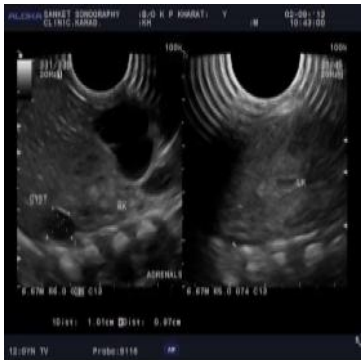

Fig.3 visceral organomegaly (cystic right adrenal gland)

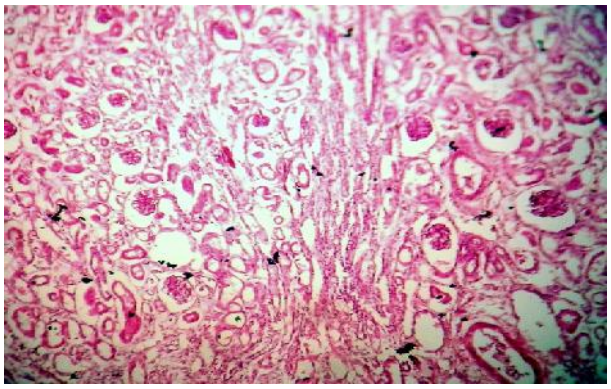

Fig.4 Microscopic sections of the kidney reveal dysorganisation of parenchyma glomerular neogenesis and increased number of immature collecting tubules (100 x H & E)

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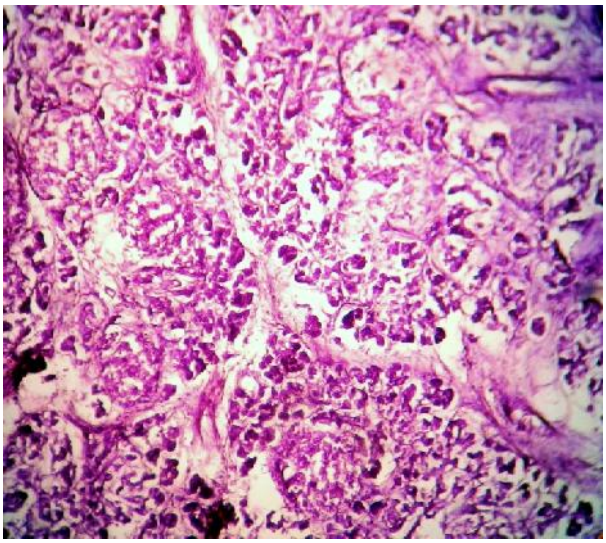

Fig.5 Microscopy sections of pancreas shows increased number of acini (400 x H & E)

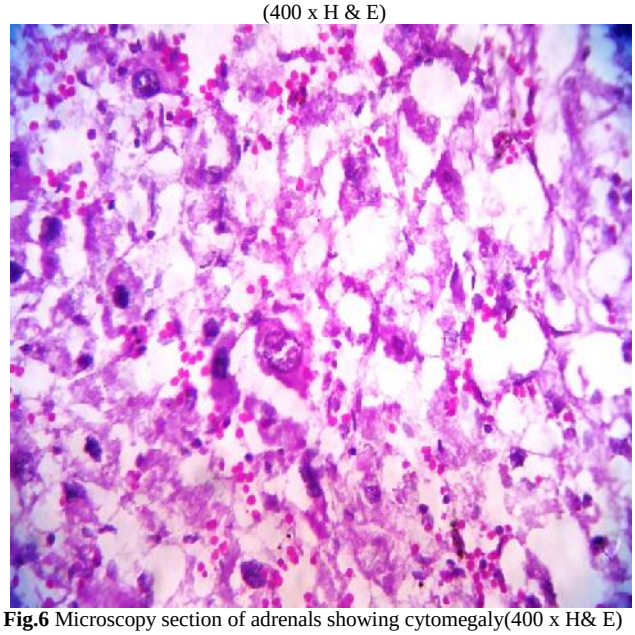

Fig.6 Microscopy section of adrenals showing cytomegaly(400 x H& E)

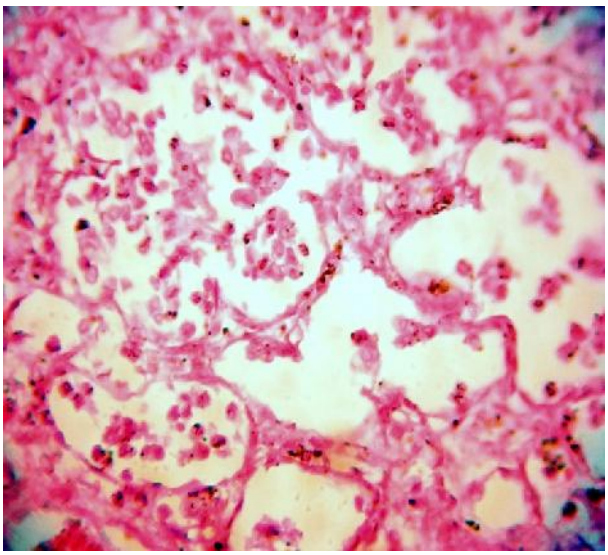

Fig.7 Lungs revealed haemorrhagic bronchopneumonia (400 x H & E)

Diagnosis: Based on morphological and autopsy findings diagnosis was given as Beckwith –Wiedemann syndrome.

DISCUSSION

Beckwith Wiedemann syndrome is a rare congenital overgrowth syndrome. The incidence is 1: 13700 live births with equal sex distribution. It was first described in 1963 by Beckwith [1].

Abnormalities involving genes on chromosome 11 are related to most cases of Beckwith Weidmann syndrome, that is abnormal DNA methylation in different areas of 11 p 15. Imprinting control regions (ICPS) controls the methylation of genes involved in normal growth[2].

These cases show increased rate of growth during later half of pregnancy and in first few years of life [3, 4]. Increased frequency of this syndrome has been observed in cases born with assisted reproduction technology[5].

These cases generally present within first 8 – 10 years of life. Abnormal growth manifest as hemihypertrophy and /or macroglossia, increased frequency of malformations like omphalocele, umbilical hernia and visceromegaly of liver, spleen, pancreas, adrenals.

Fetal adrenocortical cytomegaly is a pathognomic feature. Renal abnormalities include medullary dysplasia, nephrocalcinosis and nephrolithiasis. Rare complications include Wilms tumor, hepatoblastoma and other embryonal tumors like myosarcoma, adrenocortical carcinoma and neuroblastoma[6,7,8]. Metabolic complication of this syndrome is hypoglycemia which is seen in 30-50% of the babies related to the islet cell hyperplasia and hyperinsulinemia[9].

The diagnostic criteria for Beckwith Wiedemann proposed by Elliot and Maher are either presence of 3 major features (Macroglossia, pre-post natal growth >90 percentile and abdominal wall defects) or 2 major and 3 or more minor defects (ear lobe creases. Facial nevus flammeus, hypoglycemia, organomegaly and hemihyperthrophy)[2].

Differential diagnosis of Beckwith Wiedemann Syndrome can be Sotas syndrome, Silver Russell syndrome, fragile X syndrome, Barardinelli lip dystrophy syndrome, Marshall-Smith syndrome, Weaver- Smith syndrome [10].

Our case satisfied 2 major (macroglossia and omphalocele) and 3 minor criteria (nevus, hypoglycemia, and organomegaly) and also revealed history of assisted reproduction technology.

CONCLUSION

Beckwith Wiedemann syndrome is a rare congenital overgrowth syndrome associated with morbidity and mortality. Medical termination is not possible, as the syndrome manifests in the later half of pregnancy. Antenatal diagnosis plays crucial role for further surgical and medical management.

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