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Case Report

"LAURENCE – MOON- BIEDEL SYNDROME"- WITH END STAGE RENAL DISEASE AND FATTY LIVER.

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ABSTRACT
Laurence-Moon-Biedl syndrome is a rare, genetically heterogeneous, autosomal recessiveinherited disorder with wide variability in expression. This condition characterised by rod-cone dystrophy, postaxial polydactyly, central obesity, mental retardation, learning difficulties, hypogonadism in males and renal involvement. Renal failure is the most common cause for morbidity and mortality in such individuals. We have presented a 19 year old female patient exhibiting features of Laurence-Moon- Biedl syndrome with end stage renal disease and then the literature is reviewed**.**

Keywords: *Laurence-Moon- Biedl, Bardet Biedl Syndrome, Retinitis Pigmentosa, End Stage Renal Failure*

INTRODUCTION
Laurence-Moon- Biedl syndrome is a rare, genetically heterogeneous, autosomal recessive inherited disorder with wide variability in expression. The five cardinal features of LMBS are polydactyly, mental retardation, hypogonadism, retinitis pigmentosa, central obesity. The renal involvement is considered as 6^{th} cardinal feature.
The frequency is 1:1, 60, 000 (Beales *et al.*, 1999). With high incidence has been reported in the isolated population of New found land (1:13,000) (Green *et al.*, 1989) and Kuwait (1:17,000) (Farag and Teebi, 1989). Very few cases have been reported from India. Especially north part of Karnataka (Rajoor *et al.*, 2013). Hence we present here a case of LMBS with ESRD and grade-1 fatty liver presented to our medicine opd.

CASES
19 year old female presented to our opd with history of swelling of lower limbs, puffiness of face, fever, and reduced urine output of 15 days duration. She is a second child born to a non-consanguineous marriage, full term normal vaginal delivery with no antenatal or perinatal complications,there was a developmental delay. Learned to walkat the age of 3 ½ years, learned to speak at the age of 4 years. With family history of loss of elder brother at the age of 12 years who had polydactyly in all the limbs, with vision impairment.
On examination, patient was pale, facial puffiness, with bilateral pitting pedal oedema. Pulse rate of 72bpm, blood pressure of 138/86 mmhg. Height of 152 cm, weight of 70kg, and BMI of 31.1kg/m2.
Systemic examination reveals apex at 6^{th} intercostal place 2 cm lateral to left mid clavicular line, mild hepatomegaly.

Lab investigations

TOTAL LEUCOCYTE	7,800	TOTAL PROTEIN	6.3 g/dl	URINE ROUTINE	N
RBC	3.8millions	ALBUMIN	2.3 g/dl	Total cholesterol	146
Hb	9.4%	GLOBULIN	4 g/dl	triglycerides	77
PLATELETS	1.5 lakh	TOTAL BILIRUBIN	1.1 mg/dl	Direct HDL	30.4
ESR	32	CONJUGATED	0.7 mg/dl	LDL cholesterol	131
RBS	98mg%	UNCONGUGATED	0.4 mg/dl	VLDL	15.4
B.UREA	80mg%	AST	150 U/L		
S.CREATININE	6.5mg%	ALT	138 U/L		

Case Report

X-ray chest suggestive of cardiomegaly, ECG suggestive of left ventricular hypertrophy. Doppler echocardiography shows concentric LVH with no structural abnormalities. USG abdomen suggestive of hepatomegaly and grade-2 renal parenchymal changes, cyst at left cortico-medullary junction with normal sized kidneys. Fundus examination revealed pigmentary retinal changes in both eye. Patient also had grade-1 fatty liver with hepatomegaly and elevated liver enzymes. These changes are seen in 18.5% of markedly obese patients (Wanless and Lentz, 1990). Diagnosis of LMBS is established by clinical criteria suggested by Beales *et al.*, (1999) No genetic testing for LMBS was available at our hospital.

PRIMARY FEATURES	IN THIS CASE	SECONDARY FEATURES	IN THIS CASE
Retinitis pigmentosa	+	Speech disorder	+
Polydactyly	-	Brachydactylic	-
Obesity	+	Developmental delay	+
Learning disability	+	Polyuria/polydipsia	-
Hypogonadism	-	Ataxia	-
Renal anomalies	+	Poor co-ordination	-
		LVH	+
		Diabetes mellitus	-
		Hepatic fibrosis	-

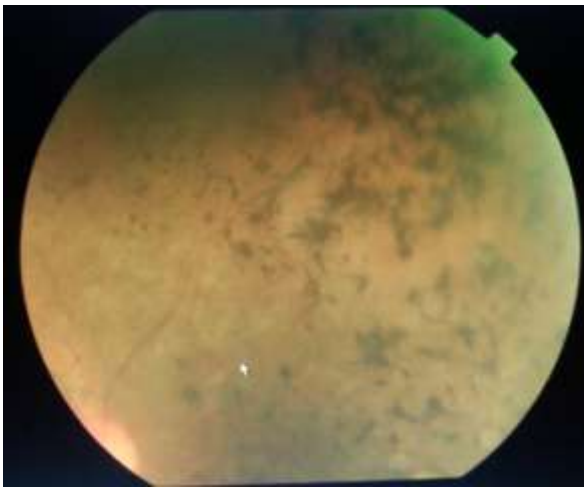

Figure 1: fundus showing retinitis pigmentosa

DISCUSSION

LMBS is a rare, autosomal recessive inherited disorder . BBS was first described by Bardet and Biedel in 1920. The exact pathogenesis is not known. Mutations in 14 genes are known to be associated with BBS: BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, TRIM32/BBS11, BBS12, MKS1/BBS13, and CEP290/BBS14 (Waters and Beales, 2011). It has been recently recognised that proteins coded for bythe BBS4, BBS6, BBS8, and BBS10 genes are expressed in the basal body of cilia, and BBS is nowregarded as one of the 'ciliopathies'. The gene products are probably involved in the signalling

Pathway in the cilia; abnormalities interfere with the normal development, resulting in the diverse pathological effects of the syndrome (Rathi *et al.*, 2007).

The five cardinal features of LMBS are polydactyly, mental retardation, hypogonadism, retinitis pigmentosa, central obesity. The renal involvement is considered as 6th cardinal feature. Due to genetic heterogeneity diagnosis of LMBS primarily relies on clinical findings & family history. This pleiotropic disorder has variable expressivity and a wide range of clinical variability observed within and between families.

Diagnosis of LMBS is established by clinical criteria suggested by Beales *et al.*, presence of 4 primaries or 4primary or 3 primary and 2 secondary features are essential for diagnosis (Beales *et al.*, 1999).

The frequency of renal involvement reported in BBS varies. Beales *et al.*, (1999) reported structural abnormalities of kidneys as renal parenchymal cysts/communicating calyceal cysts (10%), calyceal clubbing and blunting (10%), foetal lobulation (12%), scarring (12%), dysplastic kidneys (5%), unilateral agenesis (4%), renal calculi (2%), vesicoureteric reflux with pyelonephritis (9%), bladder obstruction (4%), hydronephrosis (4%), horseshoe kidney (2%), and ectopic kidney (2%). The management is mainly supportive. Rehabilitation for mental impairement, surgical correction of polydactyly, testosterone replacement therapy for hypogonadism, low fat diet, exercise for fatty liver and obesity, early screening and recognition of diabetes, hypertension, renal disease is very important. The renal failure can treated with haemodialysis, peritoneal dialysis, renal transplantation.

The purpose of this case report beside its rarity, to make reader aware of the disease entity, recognition and diagnosis, early recognition and treatment of associated comorbidities to prolong the quality and quantity of life.

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