Doi: 10.32677/IJCR.2016.v02.i01.002 **Case Report**

Rare Case Report - Brothers of Laurence Moon Biedl Syndrome with Guillain - Barre Syndrome

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ABSTRACT

Laurence Moon Biedl syndrome is an autosomal recessive disorder characterized by obesity, hypogonadism, mental retardation, polydactyly, and retinitis pigmentosa. Guillain-Barre syndrome (GBS) or acute inflammatory demyelinating poly radiculo neuropathy (AIDP) is considered to be an immunological disorder with an acute and often fulminant evolution characterised by a syndrome of rapidly progressive flaccid paralysis, aflexia and albumino-cytological dissociation in the CSF. We are here with presenting case report of brothers with Laurence *Moon Biedl syndrome (LBS) who presented with* GBS *simultaneously.*

Keywords: *Laurence Moon Biedl Syndrome; Guillain-Barre syndrome; obesity, polydactyly*

Laurence Moon (Bardet) Biedl syndrome, first defined by Bardet in 1922 is an autosomal recessive disorder characterized by structural and functional abnormalities of organs and tissues with diverse embryonic derivation [1]. The five cardinal features of the syndrome include polydactyly or syndactyly, pigmentary retinopathy, obesity, mental retardation and hypogonadism [2]. Other systemic features include brachycephaly, short stature, congenital heart block, and deafness, neurological and renal disorders [2-4].

Guillain - Barre Syndrome (GBS) is one of the commonest forms of polyneuropathy. The reported incidence rates for GBS are 1-2/1,00,000 population. The lifetime likelihood of any individual acquiring GBS is 1 in 1000. Available Indian literature indicates a peak incidence between June, July and September - October. In Western Countries, GBS is common in the fifth decade; but in India, it occurs more commonly in younger age. GBS is equally common in men and women and can occur at any age. There is a male preponderance among the hospitalized population [5].

We herewith report two cases of AIDP in brothers with Laurence Moon Biedl syndrome. To our knowledge such association in family has not been reported till date.

CASE REPORTS

These two brothers were mentally challenged, born out of consanguineous marriage, 2nd & 3rd in birth order with normal elder brother, and were admitted into medical wards in Osmania General Hospital, whose clinical presentation is given in detail below.

Case 1

18 year old male mentally challenged presented with tingling sensations in both feet, symmetrical quadriparesis of 15 days duration. This weakness remained static. There was no bowel and bladder involvement. He was non hypertensive and non diabetic. General examination revealed left eye exotropia, post axial hexadactyly (Fig 1 A, B, C), high arched palate, and bilateral gynecomastia.

His stretched penile length was 6 cm with sparse pubic hair and testicular volume of 15 ml bilaterally. He was obese with a body mass index (BMI) of 30 kg/m^2. His vitals were stable. He had mild mental retardation.

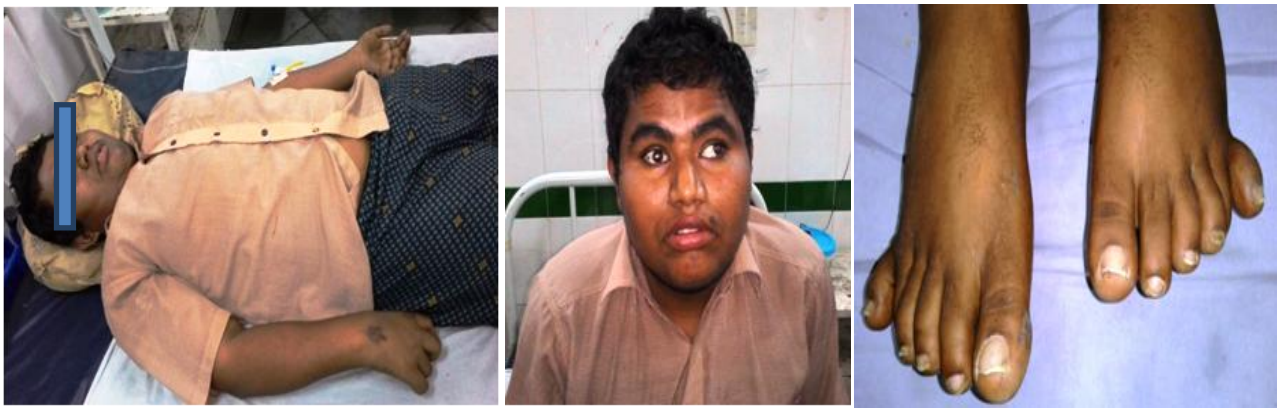

Figure 1 showing A) obesity B) left eye exotropia and C) post axial hexadactyly in first case

Motor system examination revealed symmetric flaccid quadriparesis with predominant lower limb weakness. There was no cranial nerve involvement or cerebellar involvement. Fundus examination revealed bilateral retinitis pigmentosa and macular dystrophy. Media was clear, optic disc was pale with attenuated vessels, and occasional bony spicules were present. All superficial reflexes were present with bilateral flexor plantar reflexes and absent deep tendon reflexes. Rest of the sensory system examination revealed no abnormality. There were no signs of meningeal irritation.

Complete blood picture, renal function tests, serum electrolytes, and urine examination were normal. Serum lipid profile revealed total cholesterol of 136 mg/dL, HDL cholesterol of 39 mg/dL, LDL cholesterol of 50 mg/dL, VLDL cholesterol of 47 mg/dL, and triglycerides of 236 mg/dL. His fasting blood sugars and post prandial blood sugars were within normal limits. Thyroid profile revealed a free T4 of 1.18ng/dl, and TSH of 1.87µIU/ml. ENMG was suggestive of sensory motor axonal neuropathy. Ultrasound abdomen was normal and MRI brain was normal. On 2D echocardiography, ejection fraction was 50% with global hypokinesia of left ventricle (LV), mild LV systolic dysfunction and good right ventricular function, and normal sized chambers. Colour Doppler study revealed trivial mitral and tricuspid regurgitation with no PAH.

Case 2

16 year old male who was the younger brother of previous patient presented with history of tingling sensations in foot, weakness of both lower limbs of 15 days duration which was gradually progressing. Proximal weakness was more than distal weakness. Initially, he had difficulty in getting up from squatting position, and difficulty in climbing the stairs. There was no history suggestive of cranial nerve involvement except for optic nerve, cerebellar involvement and bowel and bladder incontinence.

General examination revealed generalised obesity with BMI of 35 kg/m^2, post axial hexadactyly with finger buds in both hands (Fig 4-6), right eye exotropia, and high arched palate. Single testis was present on right side with testicular volume of 10ml and stretched penile length of 3.5cm. Patient was afebrile at the time of admission with stable vital signs. He had mild to moderate mental retardation. Fundus examination revealed bilateral retinitis pigmentosa. All other cranial nerves were normal. On motor system examination, bulk was normal; tone was decreased in all four limbs reduced power in both upper limbs (4/5) and lower limbs (3/5). All superficial reflexes were present with bilateral flexor plantar reflexes and absent deep tendon reflexes. Sensory system examination was normal and there was no obvious cerebellar involvement and no signs of meningeal irritation.

On investigations, complete blood picture, renal function tests, serum electrolytes, and urine examination were normal. His serum lipid profile showed total cholesterol of 166 mg/dl, HDL cholesterol of 42 mg/dl, LDL cholesterol of 72 mg/dl, VLDL cholesterol of 62 mg/dl, and triglycerides of 308 mg/dl. His fasting blood sugar was 88 mg/dl and post prandial blood sugar was 136

mg/dl. Thyroid profile revealed free T4 of 1.14ng/dl, and TSH of 2.89µIU/ml. Ultrasound abdomen showed grade III fatty liver and 2D echocardiography was normal, electro neuro myography (ENMG) was suggestive of sensory motor axonal neuropathy.

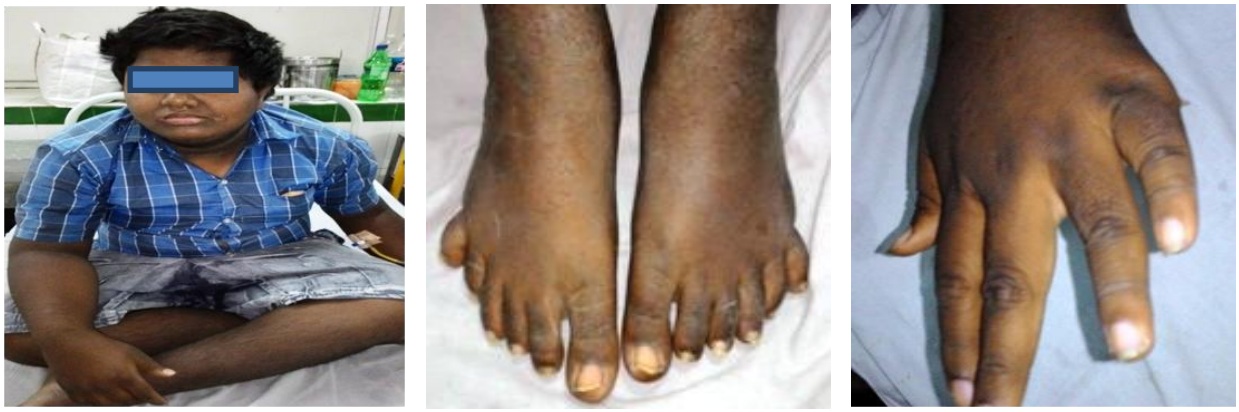

Figure 2 showing A) obesity B) post axial hexadactyly and C) finger buds in second case

Final diagnosis of Laurence Moon Biedl Syndrome with AMSAN (Acute Motor and Sensory Neuropathy) variant of GBS was made. Both the patients were treated with intravenous immune globulin (IVIg) 0.4 g/kg/day for 5 consecutive days and physiotherapy. They responded well and power improved to 4/5 in all four limbs in both individuals.

DISCUSSION

Laurenc –Moon (Bardet) Biedl is a rare genetic disorder first described by Bardet in 1920. He described three of the five cardinal features of the syndrome i.e. polydactyly, obesity and pigmentary retinopathy. Biedl in 1922 added mental deficiency and genital hypoplasia to this syndrome. In 1925, Solis-Cohen and Weiss connected this syndrome to the four patients in one family described by Laurence and Moon in 1966. Solis-Cohen and Weiss coined the name Laurence Moon Biedl syndrome. The cases were re-evaluated and reported by Hutchinson, and these members were found to have a disease characterised by tuypical pigmentary retinopathy, mental retardation, arrest of sexual development and progressive weakness leading to paraplegia [7]. Confusion exists in medical literature regarding the differences between Laurence Moon and Bardet-Biedl syndrome. Common to both are pigmentary retinal degeneration, mental retardation and hypogonadism. Spastic paraplegia is predominant feature in Laurence Moon [8].

The overlap between Bardet-Biedl syndrome and Laurence-Moon syndrome has been described before [9, 10]. We believe that the two clinical entities may represent allelic forms of the same condition and that perhaps the older term, Laurence-Moon-Bardet-Biedl syndrome should be reinstated. Moreover, perhaps a more descriptive term such as the poly dactyly-obesity-kidney-eye syndrome could be adopted.

New modified diagnostic criteria for diagnosis of LBS have been proposed according to which four primary or three primary plus two secondary features are required for diagnosing LBS [9, 10]. Primary features include rod – cone dystrophy, polydactyly, obesity, learning disabilities, hypogonadism in males, and renal anomalies. Secondary features include speech disorder/ delay, strabismus/ cataracts/ astigmatism, brachydactyly / syndactyly, developmental delay, polyuria/ polydipsia (nephrogenic diabetic insipidus), ataxia/ poor coordination / imbalance, mild spasticity (especially lower limbs), diabetes mellitus, dental crowding/ hypodontia/ small roots/ high arched palate, left ventricular hypertrophy/ congenital heart disease and hepatic fibrosis. Renal abnormalities include renal parenchymal or calyceal cysts, calyceal clubbing and blunting, scarring, renal calculi, vesicoureteric reflux with pyelonephritis, bladder obstruction, hydronephrosis, renal agenesis, horseshoe, dysplastic or ectopic kidney. In our report, both the brothers had all the major modified criteria except for renal anomalies to diagnose them as LBS.

GBS is one of the commonest forms of polyneuropathy producing a relatively symmetric a reflexic quadriparesis. The earliest description of a febrile generalized paralysis is probably that of Wardrop and ollivier in 1834. Landry in 1859 reported an acute ascending predominantly motor

paralysis with respiratory paralysis leading to death among peasants on his land. In 1916, Guillain, Barre and Strohl described a benign polyneuritis with quadriplegia with albumin cytological dissociation in Cerebro Spinal Fluid [11]. In three quarters of patients, the first neurological symptom is of paraesthesiae in the toes, less often in the finger [12].

Most of the GBS patients make a good spontaneous recovery with proper supportive treatment. GBS treatment should be initiated with either high-dose IVIg or plasmapheresis as soon after diagnosis as possible. IVIg is administered as five daily infusions for a total dose of 2 g/kg body weight [13]. The advised regimen of plasma exchange removes a total of 200 to 250 ml/kg of plasma in 4 to 6 cycles on alternate days, or over a shorter period if there is no coagulopathy. The replacement fluid is saline combined with 5% albumin [11].

CONCLUSION

GBS presenting simultaneously in family members is very rare and no case reports were there, even though the etiology is unknown in our cases. Brothers with LBS presenting with GBS may be coincidental and very rare, so we presented the above cases.

REFERENCES

1. Mc Louglin TG, Shanklin DR. Pathology of Laurence-Moon-(Bardet)-Biedl syndrome. J Pathol. 1967; 93:65-78.
2. Yamada K, Miura M, Miyayama H, Sakashita N, Kochi M, Ushio Y. Diffuse Brain stem glioma in a patient with Laurence-Moon-(Bardet)-Biedl syndrome. Pediatr Neurosurg. 2000; 33:327-7.
3. Urben SL, Baugh RF. Otolaryngologic features of Laurence-Moon-(Bardet)-Biedl syndrome. Otolaryngol Head Neck Surg. 1999; 120:571-4.
4. Ukar B, Yakut K, Kural N, Buyukasik F, Vardareli E. Renal involvement in Laurence-Moon-Bardet-Biedl syndrome: report of five cases. Pediatr Nephrol. 1997; 11:31-5.
5. AK Meena. Peripheral Neuropathy. API Text Book of Medicine, 9th edition Vol. II Jaypee Brothers Med Publishers, 2012; 1533-46.
6. Solis – Cohens, Weiss E. The Laurence Biedl syndrome: a report of four cases in one family. Am J Med Sci. 1925; 169-489.
7. Hutchihson J. Slowly progressive paraplegia and disease of choroid with defective intellect and arrested sexual development in several brothers and a sister, Arch Surg (Lond) 1900;11:118-122.
8. Laurence JZ, Moon RC. Four Cases of R.P. occurring in the same family and accompanied by general imperfections of development. Ophthalmol Rev. 1886; 2: 32-41.
9. Beales PL[1], Elcioglu N, Woolf AS, Parker D, Flinter FA. New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. J Med Genet. 1999;36:437-46.
10. Nyska M, Mozes G, Howard C, et al. Quadriparesis in the Laurence – Moon – Biedl- Bardet syndrome: case report. Paraplegia. 1991; 29: 350-4.
11. Allan H. Ropper et al; Diseases of peripheral nerve CH 46Adams and Victor's Principles of Neurology 10 TH edition,; by McGraw-Hill education. 2014; 1310-1406.
12. Micheal Donoghy; Disorders of Peripheral nerves Chapter 18th; Acute Idiopathic Polyneuritis 18.25; Brain's Disease of the Nervous System 10th Edition; Published in the United States of Oxford University Press Inc., New York. 2010; 599 – 601.
13. Anthony A. Amato, Richard J. Barohn 384th Chapter Peripheral Neuropathy, Harrison Text Book of Medicine 18th Edition, Vol II Graw Hill Publishers. 2012; 3448-50.

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