

LAURENCE MOON BIEDYL SYNDROME: A RARE CASE REPORTUdayalakshmi K.*¹, Bhavya V.¹, Hemalata A.¹, Lakshmi P.² and Dr. Ranganayakulu D.³¹Pharm-D VI Year, Department of Pharmacy Practice, Sri Padmavathi School of Pharmacy, AP, India.²Associate Professor, Department of Pharmacy Practice, Sri Padmavathi School of Pharmacy, AP, India.³Principal, Department of Pharmacy Practice, Sri Padmavathi School of Pharmacy, AP, India.***Corresponding Author: Udayalakshmi K.**

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ABSTRACT

Laurence moon Biedyl syndrome is a rare autosomal recessive condition with a wide spectrum of clinical features. The accepted major criteria for diagnosis include retinal dystrophy, obesity, Polydactyly, male hypogonadism, mental retardation and renal dysfunction. We have presented a 36 year old male patient exhibiting characteristic features of Laurence moon Biedyl syndrome and then the literature is reviewed.

KEYWORDS: Laurence, Biedyl syndrome, Polydactyly, hypogonadism.**INTRODUCTION**

Laurence – moon (Bardet) Biedyl 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 deviation.^[1] The principal manifestations are rod-cone dystrophy (Retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction. Other features not always present include hepatic fibrosis, diabetes mellitus, neurological, speech and language deficits, behavioral traits, facial dysmorphism, dental anomalies and developmental delay.^[2]

Patient generally has onset of symptoms within the first 10 years of life and most often the first complaint is poor night vision. Nystagmus is a common feature. Peripheral visual fields are constricted. Fundus changes include constricted arterioles, waxy disc pallor and peripheral pigmentary changes including pigment atrophy, Bone specular pigmentation and areas of white deposits.^[3]

EPIDEMIOLOGY

Autosomal recessive ciliopathy.

The estimated incidence is 1:160,000 in northern European population and 1:13500 in some Arab population more common in the Bedouin population of Kuwait and on the island of Newfoundland.

North American population 1:140,000.

Characterised by retinal dystrophy, renal dysfunction, post axial polydactyly, obesity, cognitive deficit and hypogonadism.

Diagnosis is based on clinical features.

CLINICAL FEATURES

- Eyes: pigmentary retinopathy, poor visual acuity, low vision or blindness.
- Nose: loss of reduced senses of, smell (anosmia).
- Hand or Foot: polydactyly or syndactyly.
- Cardiovascular system: hypertrophy of interventricular septum and left ventricle, hypertension and dilated cardiomyopathy.
- Gastrointestinal tract: fibrosis.
- Urogenital system: hypogonadism, Renal failure, Urogenital sinuses, Ectopic urethra, Uterus duplex Septate vagina, Hypoplasia of the uterus, ovaries and fallopian tube.
- Growth and developmental delay.
- Social interaction problems.
- Learning difficulties.
- Diabetes and Dyslipidaemia in some cases.



POST AXIAL POLYDACTYL - ULNAR



PRE AXIAL POLYDACTYL - RADIAL



POST AXIAL



PRE AXIAL



GENU VALGUM



GENU VARUM



RETINITIS PIGMENTOSA

PATHOPHYSIOLOGY

The dilated biochemical mechanisms that leads to this syndrome is unclear. The gene products encoded by these BBS genes, called BBS proteins, are located in the basal body and cilia of the cell. Using the round worm *C. elegans* as a model system, biologists found that BBS proteins are involved in a process called Intraflagellar transport (IFT), a bi-directional transportation activity within the cilia along the long axis of the ciliary shaft that is essential for ciliogenesis and the maintenance of cilia. Since abnormalities of cilia are known to be related to a wide range of disease symptoms including those commonly seen in BBS patients, it is now widely accepted that mutated BBS genes affect normal cilia function, which, in turns, causes BBS.

CASE REPORT

Here is a case of 36 year old male patient came to the hospital with complaints of retinitis pigmentosa, night blindness since childhood which progressed gradually over years. There was no history of consanguineous marriage between parents. Systemic examination revealed obesity, Developmental delay, and kidney problems. Fundus examination revealed that as retinitis. Cardiovascular and urinary systems were normal. Lab investigations including CBC, ESR, electrolytes and urine examinations were normal. Abdominal and echocardiogram were also normal.

DISCUSSION

The syndrome was described by Bardet Biedl in the 1920. It was later erroneously coupled with another disorder described by Laurence and Moon, and was consequently referred to as Laurence Moon-Biedl syndrome. In 1925 Solis-Cohen and Weiss connected to this syndrome 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 reported by Laurence and Moon were reevaluated and reported by Hutchinson, the members were found to have a disease characterized by typical pigmentary retinopathy, mental retardation arrest of sexual development and progressive weakness leading to paraplegia. Confusion exists in medical literature regarding the differences between Laurence moon and BardetBiedl syndrome. Common to both are pigmentary retinal degeneration, mental retardation, and hypogonadism. Spastic paraplegia is predominant feature in Laurence moon syndrome, polydactyly and obesity are predominant in BardetBiedyl syndrome. Some researchers believe that BardetBiedyl syndrome is a sub division of Laurence moon syndrome. Hence the term Laurence moon Biedl syndrome has gained universal acceptance in the world literature.

CONCLUSION

We report a typical case of Laurence Moon BardetBiedl syndrome in a male of 36 years old. He presented with

early onset blindness and other ocular features like Retinitis pigmentosa. He also showed characteristic general features of obesity. He also had kidney problems and developmental delay.

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