LAURENCE – MOON (BARDET) BIEDL SYNDROME

Tariq Qureshi; Ayub; Prof. A.R.Nasti; Mehmooda Ashai

Abstract: We report a case of Laurence Moon Biedl syndrome with Pendular nystagmus as the uncommon feature in addition to the five cardinal features.

Key words: Retinopathy, Polybactyly

INTRODUCTION
Laurence-Moon (Bardet) Biedl Syndrome, first defined by Bardet in 1922 1 is an autosomal Recessive disorder characterized by structural and functional abnormalities of organs and tissues with diverse embryonic derivation.
The five cardinal features of the syndrome include Polydactyly or Syndactyly, Pigmentary retinopathy, obeisty, Mental retardation and Hypogonadism 2. Other systemic features include Brachycephaly, short stature, congenital Heart Block, Deafness, Neurological disorders and Kidney disorders 3,4.

Patient generally has onset of symptoms within the first 10 years of life and most often the first complaint is poor night vision 5. Nystagmus is a common feature. Peripheral visual fields are constricted. Fundus changes include constricted arterioles, waxy disc pallor and peripheral pigmentary changes 6 including pigment atrophy. Bone specular pigmentation and areas of white deposits.

Unlike classic Retinitis pigmentosa unassociated with systemic disease, these patients show degenerative changes within the macula at an early stage with relentless earlier decrease in the central vision and most patients are legally blind by age 30 ERG shows diffuse photoreceptor disease 7.

CASE REPORT
A 10 year old boy reported to eye O.P.D. with complaints of poor night vision and Jerky eye movements of uncertain duration and onset, and of progressive nature. There was no history of consanguinous marriage between parents, complicated vaginal delivery or any intra-uterine infection. The other male sibling who was 2 years elder and the parents were a-symptomatic.

Systemic examination revealed polydactyly (Hexadactyly), obeisty of truncal type also involving proximal extremities, Mental retardation (I.Q <60), Hypogonadism (Pea nut size tests) and sensoneural hearing loss. Cardiovascular and urinary system were normal.

Ophthalmic examination revealed orthophoria, normal ocular
movements and pendular nystagmus in both eyes.

The unaided visual acuity in both eyes was 6/60 correctable to 6/12 with -1.50 D sph. and -0.50 D cyl. Axis 90°. Slit lamp examination of the anterior segment was normal.

Fundus examination revealed constricted arterioles, waxy disc pallor, Peripheral pigmented changes including areas of white deposits and bone spicule pigmentation. The foveal reflex was dull in appearance in both eyes.

Lab. Investigations including CBC, ESR, KFT and Urine examination were normal. Chest and abdominal Roentgenogram were normal. Serological tests were unrewarding. Abdominal ultrasound and Echocardiogram were also normal.

DISCUSSION
Laurence – Moon (Bardet ) Biedl syndrome is a rare genetic disorder first defined by Bardet in 1920 1 . He described three of the five cardinal features of the syndrome i.e; Polydactyly, Obesity and Pigmentary retinopathy.

Biedl in 1922 1 added mental deficiency and genital hypoplasia to this syndrome. In 1925 Solis-Cohen and Weiss 6 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 re-evaluated and reported by Hutchinson 7 , 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 Bardet Biedle 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 Bardet Biedl syndrome. Some researchers believe that Bardet Biedl Syndrome is a subdivision of Laurence Moon syndrome. Hence the term Laurence Moon Biedle syndrome has gained universal acceptance in the world literature.

CONCLUSION
This case illustrates several common features of Laurence Moon Bardet Biedl syndrome including the five cardinal features i.e; Polydactyly, Obesity, Mental retardation, Hypogonadism and Pigmentary retinopathy.

In addition the patient has pendular nystagmus and sensorineural hearing loss which have been reported less frequently in other series.

Reference:
8. Laurence JZ, Moon RC. Four cases of R.P. occurring in the same family and accompanied by general imperfections of development Ophthalm Rev 2:32, 1886.