Case Report

Laurence-Moon-Bardet-Biedl syndrome – A Case report with review of literature

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ABSTRACT

Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is a rare autosomal recessive (AR) disorder. It has many clinical features out of which ocular manifestations will be discussed in this article along with its ayurvedic correlation. *Acharya Sushruta* has mentioned *Adhyatmika Vyadhis* in *Sutrasthana* which is *Aadibala Pravrutta* (hereditary) *Janmabala Pravrutta* (congenital) and *Doshabala Pravrutta*. *Acharya* also elaborates on the clinical features of *Dauhrudavimaanan* (not fulfilling desires of pregnant women) in her antenatal phase in *Sharirsthana*, which has a resemblance to the clinical features of LMBBS. This case report furthermore mentions a case report of a male patient aged 20 years who visited the ophthalmic outpatient department of Shalakyatantra department in the Sane Guruji Hospital with complaints of diminished vision since childhood, diminished night vision, inability to open right eye completely, lagging of right upper eyelid, and delayed developmental milestones.

Key words: *Adhyatmika vyadhi, Dauhrudavimaanan, Doshabala pravrutta, Janmabala Pravrut*, Laurence-Moon-Bardet-Biedl syndrome

Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is a rare autosomal recessive (AR) disorder. It results from consanguineous marriage. It is characterized predominantly by hypogonadism, polydactyly, retinitis pigmentosa, obesity, and mental retardation. Primary clinical features include rod and cone dystrophy, polydactyly, central obesity, genital abnormalities, and mental retardation, often presenting as poor schooling skills. Secondary clinical features include developmental delay, speech deficit, brachydactyly/syndactyly, dental defects, ataxia, olfactory deficit, diabetes mellitus (DM), and congenital heart disease [1]. It results from consanguineous marriage [2]. The incidence of LMBBS in America and Europe varies from 1:140,000 to 1:160,000 live births. Furthermore, in Kuwait and Newfoundland, the rate is much greater, that is, an estimated rate is 1:13,500 and 1:17,500, respectively [3].

Ayurveda has explained various congenital conditions and causes behind developing anomalies in children. *Acharya Sushruta* has stated *Adhyatmika Vyadhis* in *Sutrasthana.* These are *Aadibala Pravrutta* (hereditary), *Janmabala Pravrutta* (congenital), and *Doshabala Pravrutta* (due to vitiated doshas) [4]. *Aadibala Pravrutta Vyadhis* (hereditary) occur when vitiated *Doshas* cause deformation of *Shukra* (sperm) and *Shonita* (ovum) [4]. Similarly, LMBBS is an AR disorder, which means the contribution of one affected gene from both parents can express the disorder in a child. The inheritance pattern is shown in Fig. 1.

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Janmabala pravrutta vyadhis (congenital) are *Rasakruta* (diseases due to dietary indiscretions) and *Dauhrudavimaanan* (not fulfilling desires of pregnant women). It causes *Pangutva* (lame), *Jaatandhya* (congenital blindness), *Badhirya* (deafness,) *Mukatva* (dumb), *Minminatva* (nasal twang in voice), *Vamanatwa* (dwarf) [4]. *Acharya Sushruta* also clarifies their view over *Dauhrudavimaanan* (not fulfilling the desires of pregnant women) in *Sharirsthana* by clearly stating deformities that would arise in the newborn if *Dauhruda* (desires of pregnant women) is not obeyed properly. *Acharyas* state that in 4th month of gestation, there is the formation of *Garbha Hridiya* and *Chetana*; hence, *Garbha* can feel all the sensations in this month. *Dauhrudavimaanan* (not fulfilling desires of pregnant women) in this phase could lead to *Kubja* (humpback), *Kuni* (deformed arms), *Khanja* (deformed limbs), *Jada* (obese), *Vaman* (dwarf), *Vikrutaaksha* (deformed eyes), and *Anaksha* (absence of eyes) [5].

Doshabala Pravrutta (vitiated *Doshas*) *Vyadhis* are *Sharirika* and *Mansika Vyadhis*. *Sharirik Vyadhis* are hypogonadism, polydactyly, developmental delay, speech deficit, brachydactyly/ syndactyly, dental defects, ataxia, olfactory deficit, DM, and congenital heart disease. *Mansika Vyadhis* are mental retardation, often presenting as poor schooling skills [4].

CASE PRESENTATION

A 20-year-old male patient came to the ophthalmic outpatient department of Sane Guruji Arogya Kendra, Hadapsar, Pune

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(MH, India) with complaints of diminished vision since childhood, diminished night vision, inability to open right eye completely, lagging of right upper eyelid, and delayed developmental milestones. The overall appearance of the patient is shown in Fig. 2.

From the age of 5 years, the patient had visual disturbance, which was progressive, not associated with double vision, watering, pain, or reddening of eyes. The patient had poor school performance and he was a drop-out from 7th std. He lost interest in playing games, activities, and surroundings. Surgical history of the right eye operated for ptosis repair (patient was not sure about ptosis surgery or any other ocular surgery, documents are also not available) (15 years ago).

The patient's mother had no history of any drug intake for any ailments during pregnancy. The mother also had no fever or any other medical ailments during pregnancy. The patient is born of normal, full-term vaginal delivery with no history of prolonged labor or neonatal fever, or seizures. The patient is born of a 2nd° marriage. His other sibling is absolutely normal. No other member of the family is having similar complaints.

The findings of the ocular examination of the patient are shown in Table 1. Autorefractometer and fundus readings of both eyes are shown in Tables 2 and 3, respectively.

DISCUSSION

LMBBS is a rare AR disorder. This AR disease was having bad prognosis and the patient cannot get complete relief from it. It causes hypogonadism, polydactyly, retinitis pigmentosa, obesity, mental retardation, rod and cone dystrophy, polydactyly, central obesity, genital abnormalities, and mental retardation, often presenting as poor schooling skills. Ayurveda has wisely explained about congenital anomalies as *Adhyatmika Vyadhis* in *Sutrasthana*, *i.e.*, *Aadibala Pravrutta* (hereditary), *Janmabala Pravrutta* (congenital), and *Doshabala Pravrutta* (due to vitiated doshas).

Aadibala Pravrutta Vyadhis are hereditary disorders that are caused due to deformed sperm and ovum, that is, *Shukra* and *Shonita* [4]. Thus, it clarifies that the LMBBS inheritance pattern is due to the contribution of one-one affected gene from both parents. Here, *Matruj* and *Pitruj Vikrut* factors are showing their arousal from consanguineous marriage, which is to be considered as the prime reason for the occurrence of LMBBS.

Janmabala Pravrutta Vyadhis, are the congenital disorders that occur due to improper antenatal care by the mother. These are *Rasakruta* (diseases due to dietary indiscretions) and *Dauhrudavimaanan* (not fulfilling desires of pregnant women). *Pangutva* (lame), *Jaatandhya* (congenital blindness), *Badhirya* (deafness,) *Mukatva* (dumb), *Minminatva* (nasal twang in voice), and *Vamanatwa* (dwarf) are their characteristic features [4]. *Acharya Sushruta* mentions that the development of *Garbha Hridiya* and *Chetana* come into existence in 4th month of gestation; thus, all the desires demanded by the mother are actually demanded by the fetus, and not fulfilling them can lead

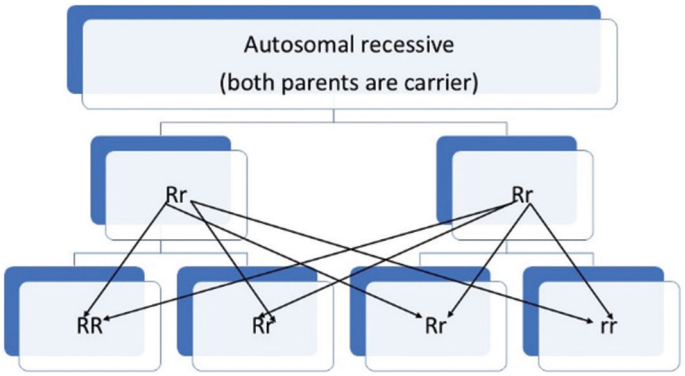

Figure 1: Inheritance pattern for Laurence-Moon-Bardet-Biedl syndrome

Figure 2: Appearance of Laurence-Moon-Bardet-Biedl syndrome patient

Table 1: Ocular examination findings of patient eyes

Ocular structures	Right eye	Left eye
Lid	Severe ptosis	N
Cornea	Clear	Clear
Conjunctiva	No congestion	No congestion
Sclera	No congestion	No congestion
Lens	Cataractous changes (nuclear)	Cataractous changes (nuclear)
Pupil	Sluggish reacting	Sluggish reacting
VA	Not cooperative	Not cooperative

Table 2: Auto refractometer readings of both eyes

Right eye	Left eye
−9.50	−9.75
−5.00	−4.25
5	160
44.25	45.50
47.00	48.00
5	165

Table 3: Fundus finding of both eyes of the patient

Fundus	Right eye	Left eye
Optic disc	Optic atrophy	Optic atrophy
Macula	Foveal reflex dull	Foveal reflex dull
Retina	Pigments noted over the retina suggestive of retinitis pigmentosa	Pigments noted over the retina suggestive of retinitis pigmentosa

to *Vikruti* due to the deterioration of *Doshas*. *Dauhrudavimaanan* (not fulfilling desires of pregnant women) in this phase could lead to *Kubja* (humpback), *Kuni* (deformed arms), *Khanja* (deformed limbs), *Jada* (obese), *Vaman* (dwarf), *Vikrutaaksha* (deformed eyes), and *Anaksha* (absence of eyes) [5]. All the mentioned deformities are likely to be seen in LMBBS patient.

Doshabala Pravrutta (vitiated *Doshas*) further lead to *Sharirika* and *Mansika Vyadhis.* As mentioned earlier, improper fulfillment of the desires of the mother causes vitiation of doshas. *Sharirik Vyadhis* mentioned in ayurvedic texts can be broadly correlated to hypogonadism, polydactyly, developmental delay, speech deficit, brachydactyly/syndactyly, dental defects, ataxia, olfactory deficit, DM, and congenital heart disease. *Mansika Vyadhis* can be correlated to mental retardation, often presenting as poor schooling skills [4].

In the above ayurvedic context, the patient in this case study has *Vikrutaaksha* (deformed eyes), *Vamantva* (dwarf), *Kuni*, *Khanja*, *Jadatva*, *Badhirya* (deafness), *Minminatva* (nasal twang in voice), *Sharirik***,** and *Mansika* manifestations. The patient was born of a 2nd° marriage, which is also considered a prime *Hetu* of disease.

Symptomatic treatment for symptomatic relief is the key treatment; hence, such sort of anomalies are better avoided. Ayurveda has wisely mentioned about the precautions that are to be taken in the antenatal phase so that there will not be any *Vikruta Garbha Nirmitti* (deformed fetus). Following *Dauhruda* (desires of pregnant women) expressed by a pregnant lady [5], the Selection of the right partner, that is, *Acharya Vagbhata* has described the qualities of the girl to be selected for marriage in detail. She should be from *Atulyagotra* (non-consanguineous), should not be suffering from any contagious diseases, must have complete body parts, should be healthy, and must possess all good qualities [6]. Other precautions included are *Upanayan Sanskara, Rajaswala Paricharya*, following *Naistiki,* and *Vaivahika Brahmacharya* [7]. Prerequisite for a healthy pregnancy as mentioned by *Acharya Charak* that, when healthy sperm passes through a healthy genital tract reaches a healthy uterus and unites with a healthy ovum, healthy conception definitely occurs [8]. Essential factors for conception of healthy progeny are *Ritu* (fertile period), *Kshetra* (well-primed endometrial bed with decidual reaction and healthy body), *Ambu* (nourishment), and *Beeja* (good quality sperm and ovum from healthy male and female) must also be taken into consideration [9]. Hence, these regimes mentioned by our *Aacharyas* could lead to healthy progeny and this sort of congenital disorders could also be wisely avoided [10].

CONCLUSION

LMBBS mentioned by modern science is briefly studied in this case report along with its ayurvedic manifestations and possible ways to avoid such progeny. Ayurveda is a holistic science and highly advanced with its unique concepts explaining the theory of genetics, embryology, rituals of cohabitation, and conditions favoring high-quality progeny. It further elaborates on what care has been taken in every aspect of the creation of high-quality offspring and how to avoid other bad circumstances related to pregnancy and anomalies in newborns.

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REFERENCES

1. Qadar LT, Ahmed ZM, Munawar M, Hasan CA, Iqbal SU. Laurence-moon-bardet-biedl syndrome with coexisting abdominal distension and positive fluid thrill: A rare manifestation reported in Karachi, Pakistan. Cureus 2019;11:e4885.
2. Kumar A, Husain A Sr., Saleem A, Khawaja UA, Virani S. Laurence-moon-bardet-biedl syndrome: A rare case with a literature review. Cureus 2020;12:e11355.
3. Katsanis N, Lupski JR, Beales PL. Exploring the molecular basis of Bardet-Biedl syndrome. Hum Mol Genet 2001;10:2293-9.
4. Priyavat S, Anantram S. Sushrut Samhita Anantram Sharma Sutrasthana (bhag 1) 24/5. Varanasi: Chaukhamba Publishing House; 2022. p. 202.
5. Priyavat S, Anantram S. Sushrut Samhita Anantram Sharma Sharisthana (bhag 2)3/18. Varanasi: Chaukhamba Publishing House; 2022. p. 35.
6. Tripathi RD. Astanga Samgraha of Srimad Vriddha Vagbhata, Sharira Sthana, 1/3. Delhi: Chaukhamba Sanskrit Pratisthana; 2006.
7. Harijan RH, Yadav CR, Singh BP. Role of ayurveda in the prevention of genetic disorders. AYUSHDHARA 2023;10:29-35.
8. Kasinath S. Vidyotinivyakhya, Charak Samhita, Sharir 8/17. Varanasi: Chaukhamba Bharati Academy; 2005. p. 825.
9. Priyavat S, Anantram S. Sushrut Samhita Anantram Sharma Sharisthana (bhag 2)2/32. Varanasi: Chaukhamba Publishing House; 2022. p. 21.
10. Dhiman K, Kumar A, Dhiman KS. Shad garbhakara bhavas vis-a-vis congenital and genetic disorders. Ayu 2010;31:175-84.

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