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The second case of Bardet-Biedl syndrome from Iraq: An educational article and expert opinion

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Abstract

Bardet-Biedl syndrome and Laurence-Moon syndrome are rare autosomal recessive disorders with a similar phenotype. They are characterized by the gradual development of phenotype which includes cone-rod retinal dystrophy, obesity, and hypogonadism in males. Other associated abnormalities include learning disability and renal abnormalities. The two conditions are differentiated clinically by the presence of spasticity and the absence of polydactyly in Laurence- Moon syndrome. In 2002, we reported the first patient with Bardet-Biedl syndrome. The boy had polydactyly, obesity, retinal degeneration, and was hospitalized at the University Hospital in Al-Kadhimiyia because of the development of chronic renal failure. The case of a 9-year-old obese Kurdish girl who had polydactyly of the hands and feet associated with reduced visual acuity is described. A 9-year-old obese Kurdish girl had polydactyly of the hand and feet associated with reduced visual acuity that was attributed to myopia which was partially corrected with eye glasses. The parents were unrelated, and her younger 5-year sister had polydactyly of the feet only. No other family member known to have polydactyly. Both sisters had their extra- digits removed surgically. The girl was studying at fourth grade primary school and the parents reported no learning disability. Fundoscopy showed normal optic disc and periphery of the retina. The presence of retinitis pigmentosa was excluded. However, the foveal reflex was reported to be abnormal by the oophthalmologist. Electroretinography showed evidence of cone-rod dystrophy. The second case of Bardet-Biedl syndrome from Iraq is reported. The current evidence-based expert opinion suggests the use of long tem supplementation of citicoline, co-enzyme Q10 and lutein with hope of retarding the progression to blindness.

Keywords: *bardet-biedl syndrome; Iraq; evidence-based therapies; expert opinion*

Introduction

Bardet-Biedl syndrome and Laurence-Moon syndrome are rare autosomal recessive disorders with a similar phenotype. They are characterized by the gradual development of phenotype which includes cone-rod retinal dystrophy, obesity, and hypogonadism in males. Other associated abnormalities include learning disability and renal abnormalities.

The two conditions are differentiated clinically by the presence of spasticity and the absence of polydactyly in Laurence-Moon syndrome [1-6].

In 2002, we reported the first patient with Bardet-Biedl syndrome. The boy had polydactyly, obesity, retinal degeneration, and was hospitalized at the University Hospital in Al-Kadhimiyia because of the development of chronic renal failure.

Patients and methods

The case of a 9-year-old obese Kurdish girl who had polydactyly of the hands and feet associated with reduced visual acuity is described.

Results

A 9-year-old obese Kurdish girl had polydactyly of the hand and feet associated with reduced visual acuity that was attributed to myopia which was partially corrected with eye glasses. The parents were unrelated, and her younger 5-year sister had polydactyly of the feet only. No other family member

known to have polydactyly. Both sisters had their extra-digits removed surgically.

The girl was studying at fourth grade primary school and the parents reported no learning disability.

Urinalysis and renal functions showed no abnormality.

Ophthalmological examination was performed to confirm the suspected diagnosis of Laurence-Moon-Bardet-Biedl syndrome.

Fundoscopy showed normal optic disc and periphery of the retina. The presence of retinitis pigmentosa was excluded. However, the foveal reflex was reported to be abnormal by the oophthalmologist.

Therefore, electroretinography was performed bases on the early emphasis of Prosperi and colleagues on the value of electroretinography in the early diagnosis of the Laurence-Moon-Bardet-Biedl syndrome by showing evidence of retinal degeneration that is not always identified by ophthalmoscopy during childhood [5].

Electroretinography showed evidence of cone-rod dystrophy.

The parents were very anxious because the ophthalmologist informed them that the child will become blind in the future. They requested the use of possible preventive therapies to preserve her vision. Therefore, safe evidences-based therapies were recommended.

We recommended a preventive therapeutic approach including citicoline (An initial one-month course of intramuscular citicoline followed long-term citicoline eye drops or oral citicoline), long term supplementation of co-enzyme Q10 and lutein based on the evidence provided by Mao and colleagues (2016), Parravano et al (2020), Zhang et al (2017), and Bahrami colleagues (2006) [7,8,9,10].

Discussion

Bardet-Biedl syndrome and Laurence-Moon syndrome are rare autosomal recessive disorders with a similar phenotype.

Laurence-Moon syndrome was first described in 1866 by John Zachariah Laurence (Figure-1A) and Robert Charles Moon (Figure-1B). They described 4 patients from the same family who had developmental delay associated with retinitis pigmentosa.

Bardet-Biedl syndrome was named after French and Hungarian physicians who suggested that the condition is a distinct clinical entity during the 1920s.

Georges Louis Bardet was a French medical student at the University of Paris in 1920. He reviewed in his medical degree thesis the previously reported cases associated with obesity, retinitis pigmentosa and polydactyly including the cases of Darier in 1887, De Cyon in 1889, Marguerite Catt in 1914, and other cases. Marguerite Catt described a 39-year patient with polydactyly, mental retardation, obesity, retinitis pigmentosa, hypogonadism.

In addition, Bardet described a case of an obese child with hexadactyly and retinitis pigmentosa, and called the syndrome adiposo-genital dystrophy [2].

In 1922, Arthur Bield (Figure-2) described the condition in two sisters.

Fig 1A: John Zachariah Laurence (1829-1870), an English ophthalmologist

Fig 1B: Robert Charles Moon (1844-1914), an English ophthalmologist

Fig 2: Arthur Biedl (October 4, 1869-August 26, 1933) was a Hungarian pathologist born in what today is Comloșu Mic, Romania

In 2005, Susan J Moore from Canada and her research team reported a clinical genetic and

epidemiology study of Bardet-Biedl syndrome, and emphasized that Bardet-Biedl syndrome and Laurence-Moon syndrome are two disorders with a similar phenotype. They are characterized by the gradual development of phenotype which includes cone-rod retinal dystrophy, obesity, and hypogonadism in males. Other associated abnormalities include learning disability and renal abnormalities. The two conditions are differentiated clinically by the presence of spasticity and the absence of polydactyly in Laurence-Moon syndrome [4].

For this patient, we recommended a preventive therapeutic approach including citicoline (An initial one-month course of intramuscular citicoline followed long-term citicoline eye drops or oral citicoline), long term supplementation of co-enzyme Q10 and lutein based on the evidence provided by Mao and colleagues (2016), Parravano et al (2020), Zhang et al (2017), and Bahrami colleagues (2006) [7,8,9,10].

Mao and colleagues (2016) reported an experimental study which showed that citicoline can help in retarding myopia progression in guinea pigs [7[.

Parravano et al (2020) reported a study which showed that citicoline and vitamin B_{12} eye drops have a protective effect against functional impairment and neuro-retinal degeneration in mild diabetic retinopathy [8].

Zhang et al (2017) reviewed the literature and emphasized the presence of a good evidence suggesting a therapeutic potential of Co-enzyme Q10 in a variety if retinal diseases including retinitis pigmentosa. They attributed the beneficial therapeutic effect to inhibition of reactive oxygen species generation and protecting neuro-retinal cells from oxidative damage [9].

Bahrami colleagues (2006) reported a placebo-controlled study which showed that lutein supplementation can has a beneficial effect in retinitis pigmentosa and may improve visual field and also possibly improve visual acuity [10].

Conclusion

The second case of Bardet-Biedl syndrome from Iraq is reported. The current evidence-based expert opinion suggests the use of long tem supplementation of citicoline, co-enzyme Q10 and lutein with hope of retarding the progression to blindness.

Conflict of interest: None.

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