Bardet Biedl syndrome and retinopathy pigmentosa: about a case

Abstract:

Bardet-Biedl syndrome (BBS) is a rare poly-malformative syndrome of autosomal recessive inheritance. It combines retinal dystrophy, obesity, hypogonadism and often moderate mental retardation. We report the case of a 23-year-old patient referred to the nephrology department for progressive bilateral visual acuity loss.

Key words: retinopathy pigmentosa- poly-malformative syndrome- recessive transmission.

Introduction :

Bardet-Biedl syndrome is a rare multi-organ genetic disorder classified as a ciliopathy. The disease is inherited autosomal recessively, and is associated with retinal dystrophy, obesity, polydactyly, hypogonadism, mental retardation and renal impairment.

Clinical case:

This is a 23-year-old patient from a second-degree consanguineous marriage, with a history of visual impairment since childhood, obesity, intellectual difficulties, operated polydactyly, chronic renal failure at the hemodialysis stage (3 sessions per week), with a history of deafness, and gait abnormalities.

Ophthalmological examination was difficult because of nystagmus and photophobia, visual acuity was at near finger count in both eyes, anterior segment and tone examination was unremarkable, fundus showed bilateral retinitis pigmentosa (figure 1).

Macular OCT objective bilateral degenerative macular edema (figure2).

Discussion:

Bardet-Biedl syndrome is a ciliopathy due to a mutation identified on different chromosomes encoding 16 proteins, first described in 1920 by Bardet and then by Biedl in 1922. The mode of transmission is usually autosomal recessive. It leads to multivisceral damage.

The diagnosis of BBS is based on the identification of major clinical criteria, confirmed by the identification of mutations in one of the causative genes.

The diagnosis is based on the presence of at least four of the following criteria, including at least one of the first two:

- The ophthalmological features of BBS are essentially characterized by mixed cone-rod dystrophy. It appears later in life. According to Bonneau, 15% of subjects have an abnormal fundus between the ages of five and ten (1). The phenotype of retinal damage is characterized by the constant presence of osteoblasts (3).
- Polydactyly >75%,
- Obesity >80%: severe early onset after the first year of life, when there is often no obesity in the family
- Kidney abnormalities, sometimes of antenatal onset 50%, major cause of mortality in BBS patients.

- Intellectual disability and/or behavioural disorders,
- Hypogonadism (males) or genitourinary malformations 60% (2)
- Some children have gait abnormalities and coordination difficulties, as in our case.

There is currently no specific treatment to cure BBS.

Treatment is symptomatic and preventive (4,5).

- Speech therapy, which should be started at the first signs of language disorders
- A balanced diet.
- Orthoptics to combat the decline in peripheral vision.
- Psychomotricity to combat coordination disorders.
- Psychological support.

Conclusion:

Bardet Biedl syndrome is a rare and serious disease, whose functional prognosis is linked to visual impairment or blindness, mental retardation and obesity.

The vital prognosis is linked to renal impairment, which must be systematically detected.

Diagnosis is based on clinical criteria and confirmed by genetic counseling.

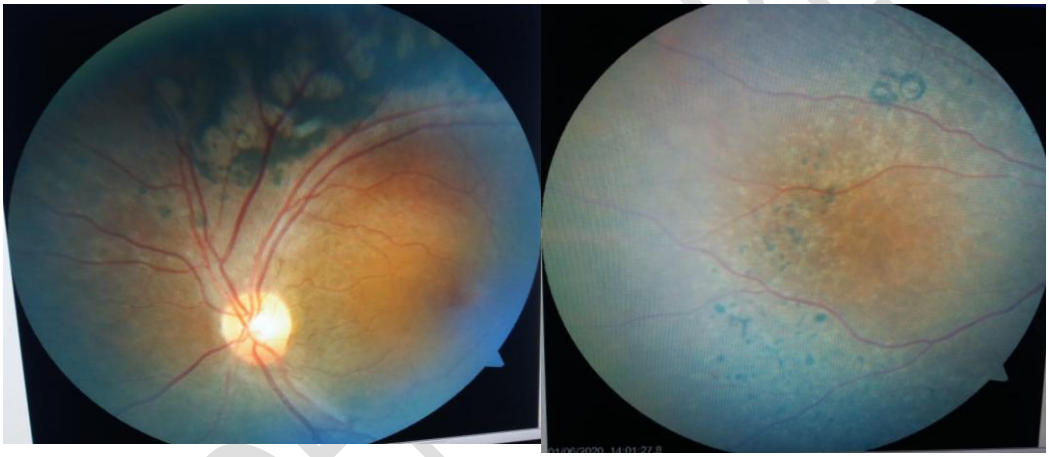

Figure 1: retinophoto showing pigmentary retinopathy bilaterally

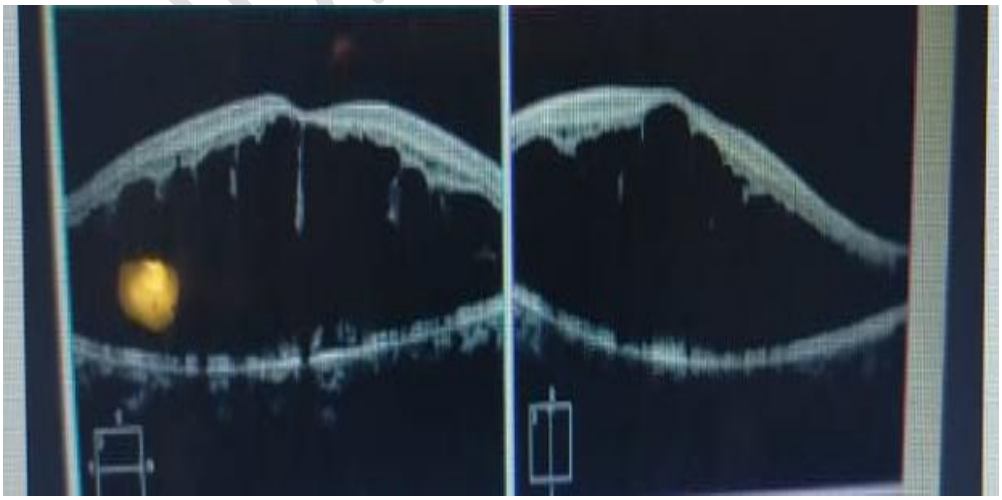

Figure2: Macular OCT showing macular edema

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