

## AUTISM AND FACIAL SYMPTOMATOLOGY IN BARDET BIEDL SD. CASE REPORT

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### *Abstract*

Autism is a mental transtorn that can be showed in many diseases and syndromes as Bardet Biedl sd(BBs) that we present. We make an introduction about autism and this syndrome, in general, then, both, in particular, then we present the case of this patient and finally the psiquiatric manifestations and facial of this child that are in the autism espectre. As a result of medical treatment, he is better.

### **Keywords:**

*Bardet Biedl sd, autism, DSM-V, facial abnormalities*

### **Introduction**

Autism is a mental trastorn with diagnostic criteria in DSM V(1) as: persistent deficits in social communication and social interaction across multiple contexts, restricted, repetitive patterns of behavior, interests, or activities; symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life); symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning, these disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay; intellectual disability and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level.

BBs is a rare genetic disease type that produces very different effects on the (multisystem) organ systems. Mainly characterized by manifestations of obesity, retinitis pigmentosa, polydactyly, mental retardation, hypogonadism,renal failure.

So far, 12 genes have been involved: (BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8,BBS9,BBS10,BBS11,BBS12).

The syndrome is familiar and is transmitted as an autosomal recessive. The locus on chromosome 3 appears linked to polydactyly of the four members, whereas chromosome 15 is associated with morbid obesity and is mostly confined to the hands, and chromosome 16 represents the "flat" form.

### Clinical Manifestations(2)Fig1.

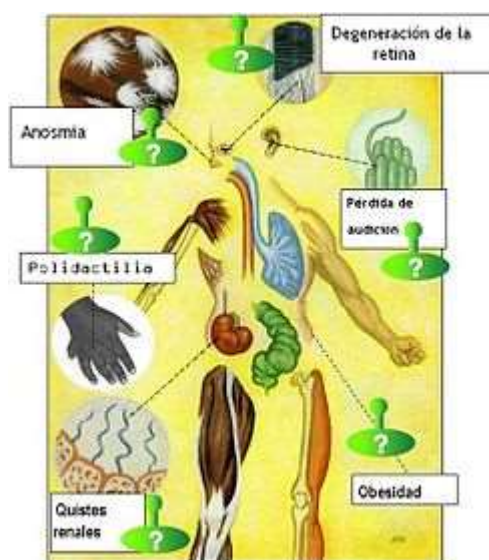
- Eyes: retinitis pigmentosa, poor visual acuity, visual impairment and / or blindness caused by problems in the transport mechanism photoreceptors retina.
- Nose: Loss or reduction of sense of smell (anosmia). Some patients report a sense of smell-sharper.
- Hands and feet: polydactyly (supernumerary fingers) or syndactyly (fusion of fingers).
- Cardiovascular system: hypertrophy of the interventricular septum and the left ventricle,dilated-cardiomyopathy.
- Gastrointestinal: fibrosis.
- Genitourinary system: hypogonadism, renal failure, abnormalities of the urogenital sinus (persistent cloaca), ectopic urethra, double uterus, septate vagina and hypoplasia uterus,ovaries,fallopian-tubes.
- Mental-growth-retardation.
- Conduct and capabilities: we have identified several problems of socialization and social interaction associated with BBs. Some authors refer to them as a form of mild autism.
- Heat sensitivity and mechanosensitivity. A 2007 study determined that genetic alterations in proteins multiprotein complex called BBSoma produce defects in the function of peripheral sensors on the species humana.
- Other symptoms: obesity, probably related to a decrease in that normally indicates satiety sensory function, facial

dysmorphism as deep set eyes, hypertelorism with downward slanting palpebral fissures, a flat nasal bridge with anteverted nares and prominent nasolabial folds, a long philtrum, and a thin upper lip. Many BBS patients have a prominent forehead, while adult males show early balding, giving them the appearance of being much older than their chronological age and dental anomalies in 27% as crowding, malocclusion/ mild micrognathia, enamel hypoplasia, discolouration and yellow discoloration, high arched palate(88%)(3,4,5,6,7).

Treatment is mainly symptomatic(8), trying each of the complications that arise during the course of the disease.

### Case report

We present a boy of 11 years old with BBS diagnosed by its neuropediatrician who appear in Child and Adolescent Psychiatry consultation with a symptoms of alteration as language delay and progressive evidence of mental retardation and traits of impulsivity. Highlights repetitive behaviors, lack of symbolic play, intolerance to changes and difficulties in social interaction compatible with ASD. and self-injurious behavior. Familiar history are Unrelated healthy parents. Mother schizoaffective disorder diagnosis.



*Fig1. Symptomatology*

Younger sister of age with psychomotor retardation, morbid obesity, retinal atrophy and asymmetry TMANO with left renal ectasia grade II/ III.

Personal history are: pregnancy controlled uneventful; term delivery, cesarean delivery no progression; PN: 3900Kgr; normal neonatal period; normal metabolic screening; food intolerances regulated without; psychomotor delay in walking at 18 months. Exploring present syndactyly between 3rd and 4th finger of both feet, prepubertal Tanner, acromimia, micropenis 3-4cm urethral meatus centered left cryptorchidism with atrophic testis right initially and finally no-palpable, smallteeth, obesity intense high, insulin resistance with impaired fasting glucose, hypertriglyceridemia, lobulations fetal kidneys, subcapsular opacity connictalopia, non-progressive chronic encephalopathy and subclinical hypothyroidism.

Investigations present EEG pattern consistent with diffuse encephalopathy with slowing of activity base. MRI brain: Normal. Normal karyotype. Patron normal methylation in Prader Willi critical region. In a second request for karyotype made from consultation and after-liaison with clinical genetics and pediatric endocrinology writing a review is requested looking compatible pattern with BBs.

### Results and discussion

In this case the treatment used for behavioral control is injectable paliperidone, oxcarbazepine and clonazepam getting a good control of aggression and stereotypies. As a doctor added thyroxin treatment.

Initially attempted dispense transdermal testosterone patches not being positive for behavioral deterioration because of the increasing of aggression.



Surgery treatment of facial abnormalities is questioned by many surgeons as cosmetic but we think that many behavioral trastorn is increased by rejection of society because of facial problems so if this patient, with normal or not very low intellectual coefficient(9), be operated of face, can avoid the stigma and could be integrated in society.

### Conclusion

In cases of autism should assess the possible clinical symptomatology companion to rule that is a child autism as if BBs or some secondary disease and facial abnormalities are closely related with mental secondary diseases so maxillofacial surgery must work next to psychiatry and psychology in many mental and facial trastorns.

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