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BARDET-BIEDL SYNDROME: A CASE SERIES

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Introduction:

Bardet-Biedl syndrome is a rare autosomal recessive disease, characterized by rod-cone dystrophy, truncal obesity, left foot polydactyly, cognitive impairment, male hypogonadotrophic hypogonadism, female genitourinary malformations, and renal abnormalities.
The authors present 3 cases of Bardet-Biedl syndrome with diagnosis in pediatric age.

Case 1:

- 3-year-old female with family history of consanguinity
- Referred to outpatient clinic at 16 months due to obesity+dysmorphic features+developmental delay
- Diagnosis at age of 22 months

Primary features	
Rod-cone dystrophy	
Polydactyly (corrected at 8 months age)	X
Obesity	X
Renal anomalies	X
Hypogonadism (male sex)	
Learning difficulties	X

+ Bilateral foot brachydactyly

Homozygous mutation in BBS7 gene

- Nutritional support
- Occupational and speech therapy

Case 2:

- 12-year-old female with family history of consanguinity
- Referred to outpatient clinic at 2 years due to obesity+dysmorphic features+developmental delay+ post-axial polydactyly
- Diagnosis at age of 2 years

Primary features	
Rod-cone Dystrophy (diagnosis at age of 7 years)	X
Polydactyly	X
Obesity	X
Renal anomalies	
Hypogonadism (male sex)	
Learning difficulties	X

+ Clinodactyly of the 5th finger

Neuroblastoma at 6 months (ressected at age of 11 months)

Positive genetic testing

System of visual amplification in daily activities

Case 3:

- 13-year-old male with no family history of consanguinity
- Referred to outpatient clinic in first year of life due to obesity+ post-axial polydactyly+ developmental delay
- Diagnosis at age < 5 years

Primary features	
Rod-cone dystrophy	X
Polydactyly	X
Obesity	X
Renal anomalies	(X)
Hypogonadism (male sex)	X
Learning difficulties	X

+ Syndactyly of both hands and feet

+Nephrocalcinosis (and suspected medullary sponge kidney)

Mutation in BBS1 gene

- Adapted school curriculum
- Speech therapy

Conclusion: As shown by our case series, Bardet-Biedl syndrome has a significant interfamilial variation. There is currently no cure for Bardet-Biedl syndrome; patients require a symptomatic and preventive approach and a close multidisciplinary follow-up.

Source: Forsythe E., Beales PL. Bardet-Biedly syndrome. European Journal of Human Genetics (2013) 21, 8-13.