



PP-RH-03

METABOLIC CHARACTERISTICS OF PATIENTS WITH POLYCYSTIC OVARY SYNDROME IN A PERUVIAN HOSPITAL

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OBJECTIVES

To describe the metabolic characteristics of patients with PCOS in the endocrinology service of a Peruvian hospital.

METHODOLOGY

Descriptive study that evaluated the clinical and metabolic characteristics of patients with PCOS in the endocrinology service of the Edgardo Rebagliati Martins National Hospital, Lima - Peru.

RESULTS

We evaluated 100 consecutive women; age 27.2 ± 7.3 years; 4% and 7% had DM2 and hypertension, respectively. 66% had a family history of DM2, 37% hypertension, 38% obesity, and 9% early cardiovascular disease. Twelve women had macrosomic children. Their BMI: 27.9 ± 6.4 g/m²; 68% had a BMI >25 kg/m², 47% had a waist circumference > 88 cm, and 37% had acanthosis nigricans.

Fasting blood glucose: 91.9 ± 23.4 (range: 68–214 mg/dL); fasting insulin and 2 hours post-load: 15.8 ± 11.2 (range: 2.7–77.4) and 59.3 ± 59.7 (range: 8.4–400), respectively. Some 15% presented with basal hyperglycemia >100 mg/dL and 42% had basal hyperinsulinemia >12.5 uIU/ml.

Forty six percent had HOMA-IR >2.5; 66% had QUICKI <0.357 and 22% had a Glucose/Insulin ratio <4.5. HOMA-%B fluctuated between 30.9 and 928.8%; 28% had HOMA-%B <100% and 23% HOMA-%B >300%. 62% had OGIS method <405 and 32% ISI-composite <3.

HDLc and triglyceride concentrations were 45 ± 11.4 (range: 20–79 mg/dL) and 123 ± 75.3 (32–390 mg/dL), respectively. 54% had HDLc <50 mg/dL and 25% had triglycerides >150 mg/dL.

CONCLUSION

Increased weight, positive family history and metabolic alterations are frequent in PCOS. The different surrogates for establishing insulin resistance, both fasting and during oral glucose load, are variable. It is important to establish the PCOS phenotype present in each patient.

PP-RH-04

PRIMARY AMENORRHEA IN AN ADOLESCENT FEMALE AS A PRESENTING FEATURE OF BOUCHER-NEUHÄUSER SYNDROME

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BACKGROUND

Primary amenorrhea has multiple aetiologies, encompassing disorders of ovaries, outflow-tract, pituitary/hypothalamus and genetic conditions. Hypogonadotropic hypogonadism refers to secondary hypogonadism due to deficiency in gonadotrophins, resulting in lack of estradiol stimulation, commonly due to pituitary or hypothalamus lesions, but can be due to genetic causes.

CASE

We describe a 16-year-old female who presented with primary amenorrhea and lack of secondary sexual characteristics, with no underlying medical conditions. Her sister and mother attained menarche at age 14. On examination, her BMI was 25 kg/m² and height was 151 cm (mid-parental height 155 cm). She had no syndromic features, no anosmia, and no hirsutism or features of virilization, and no visual or neurological abnormalities. External genitalia was consistent with infantile labia with intact introitus. Tanner-staging for her breast was 2/5 and 1/5 for genitalia and pubic hair.

Her hormonal profile showed hypogonadotropic hypogonadism, with undetectable estradiol (<36.7 pmol/L) and low gonadotrophin levels: Luteinizing-hormone 1.14 IU/L (2.4–12.6) and Follicular-stimulating-hormone 2.61 IU/L (3.5–12.5)]. Her bone age was delayed at 14 years, compared to a chronological age of 16. Karyotyping was 46,XX. Pelvic MRI showed hypoplastic uterus with normal vagina and ovaries. A pituitary MRI revealed a normal pituitary gland. Genetic testing confirmed 2 variants of PNPLA6 gene in keeping with Boucher-Neuhäuser syndrome

CONCLUSION

Boucher-Neuhäuser syndrome (BNS) is a rare genetic disorder characterized by cerebellar ataxia, chorioretinal dystrophy, and hypogonadism. We illustrate an interesting case presenting with isolated hypogonadotropic hypogonadism with genetic screen suggestive of BNS, underlying the importance of considering genetic causes of primary amenorrhea even when presentation is in adolescence.