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P-625  In vitro maturation of oocytes: a breakthrough for treating infertility in inactivating mutation of the luteinizing hormone/choriogonadotropin receptor

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Study question: How can pregnancy and live birth be achieved in patients with inactivating homozygous luteinizing hormone/choriogonadotropin receptor (LHCGR) mutation?

Summary answer: We report the first live birth after in vitro maturation (IVM) in a patient with a novel homozygous inactivating LHCGR mutation (exon 6, c.470 A>G).

What is known already: LH plays a fundamental role in female reproductive physiology and is responsible for steroidogenesis, oocyte maturation, ovulation and subsequent progesterone production by the corpus luteum. LH binds to the LHCGR located on the membrane of theca cells and mature granulosa cells. Inactivating mutations of the LHCGR lead to the impossibility to obtain final oocyte maturation both during natural cycles and after ovarian stimulation for IVM fertilization purposes. Therefore, egg donation represents the only option for treating their infertility.

Study design, size, duration: A case report.

Participants/materials, setting, methods: A 35 year-old nulliparous patient referred to our university hospital for primary infertility.

Main results and the role of chance: The 35-year-old nulliparous patient presented with primary spaniomenorrhea but timely and spontaneous onset of secondary sexual characteristics. Serum LH levels were high (ranging from 15 to 30 IU/L) and to a lesser extent so were FSH levels. The ovarian reserve was normal for age, as assessed by serum AMH levels and ultrasound. There was no argument for polycystic ovarian syndrome, 21-hydroxylase deficiency, Cushing’s syndrome or hyperprolactinemia. Two previous attempts of controlled ovarian stimulation with gonadotropins and ovulation trigger with hCG and GnRH-agonist trigger had failed, resulting in low estradiol levels despite correct follicular growth on ultrasound and absence of ovulation after trigger. However, genetic analysis identified a novel homozygous inactivating LHCGR mutation (exon 6, c.470A>G) which had never been described previously. IVM was performed. A total of 7 oocytes were obtained after IVM, resulting in 4 Day 3 embryos. All embryos were frozen. Subsequently, 2 Day 3 embryos were replaced after endometrial preparation by hormone replacement therapy. The patient became pregnant and gave birth to a healthy baby. Two Day 3 embryos remain.

Limitations, reasons for caution: By definition, a case-report requires further studies to confirm our findings.

Wider implications of the findings: We describe a novel inactivating LHCGR mutation with subsequent live birth after IVM. As a growing number of gonadotropin receptor mutations are identified, this successfully achieved live birth places IVM as the only reliable option for these patients to conceive with their own eggs.

Trial registration number: Not applicable.