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Biometra seminars

May 3, 2022 – 1:30 PM
AULA B, LITA Segrate

CONGENITAL HYPOGONADOTROPIC HYPOGONADISM: A COMPLEX AND RARE HUMAN NEUROENDOCRINOLOGICAL DISORDER DUE TO A DYSFUNCTION OF THE GNRH-SECRETING NEURONS

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Human fertility is completely dependent upon the proper development and physiology of a small number of hypothalamic neurons that secrete the neuro-hormone GnRH. A failure of GnRH neurons to develop or integrate properly leads to Congenital Hypogonadotropic Hypogonadism (CHH), which is characterized by congenital GnRH deficiency, absent or delayed puberty and consequent infertility. Different aspects of CHH are still far from being fully identified and understood, starting from its complex etiopathogenesis to several aspects concerning the patients' clinical managements. Indeed, animal models and genetic-linkage studies uncovered several molecular regulators of GnRH neuron development, although around half of the CHH cases are still classified as idiopathic and a genetic cause not detectable. On the other hand, unsolved questions regarding the wide spectrum of the clinical phenotypes, the diagnostic and therapeutic approaches of CHH patients are present and largely debated.

The present talk will illustrate the contribution of our group in this field either in terms of a better comprehension of the etiopathology mechanisms underlying CHH or a more detailed clinical characterization and a more tailored management of these patients.

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