



## TITLE: Whole Exome Sequencing of Genes Involved in Disruption of Hypothalamo-Pituitary-Testicular Axis Leading to Failed Puberty

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### ABSTRACT (up to 300 words)

Of 598 genes involved in multiple processes of the hypothalamo-pituitary-testicular (HPT) axis development, mutations in only 75 genes were shown to cause delayed puberty. We selected six male patients with failed puberty, manifested as absent signs of puberty by 18 years of age, for whole exome sequencing of genomic DNA with subsequent bioinformatics analysis and confirmation of selected variants by Sanger sequencing. Genes in which plausibly pathogenic non-synonymous variants were identified were divided into group A (previously reported to cause delayed puberty), group B (expressed in the HPT-axis but no mutations therein were reported to cause delayed puberty) and group C (not reported previously to be connected with HPT-axis). Our results identified variants in genes involved in GnRH neuron differentiation (2 in group A, 1 in group C), GnRH neuron migration (2 each in groups A and C), development of GnRH neural connections with supra-hypothalamic and hypothalamic neurons (2 each in groups A and C), neuron homeostasis (1 in group C), molecules regulating GnRH neuron activity (2 each in groups B and C), receptors/ proteins expressed on GnRH neurons (1 in group B), signaling molecules (3 in group C), GnRH synthesis (1 in group B), gonadotropins production and release (1 each in groups A, B, and C) and action of the steroid hormone (1 in group A). In conclusion, non-synonymous variants were identified in 16 genes of the HPT-axis, which comprised 4 in group A that

contains genes previously reported to cause delayed puberty, 4 in group B that are expressed along the HPT-axis but no mutations therein were reported previously to cause delayed puberty and 8 in group C that contains novel candidate genes, suggesting wider genetic causes of failed male puberty.

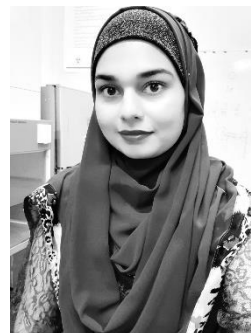
### BIOGRAPHY (up to 200 words)

Dr. Maleeha Akram has completed Ph.D. in her early 30s from the Department of Zoology, Wildlife and Fisheries, Pir Mehr Ali Shah Arid Agriculture University Rawalpindi, Pakistan. She has two research articles and one review paper published in different journals of international repute. She also has two conference papers presented in European Congress of Endocrinology out of her Ph.D. work. In total, she has seven full length research papers and twenty-nine conference papers in her credit with an impact factor of 12.28.

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