

PRESS RELEASE

Faulty gene can delay or block puberty

Scientists have identified a new gene that may influence the timing of puberty, according to research presented today at the Society for Endocrinology annual BES conference. Until now, very little has been known about the genetic control of puberty.

More than 4% of adolescents suffer from early or late-onset puberty, which is associated with health problems including obesity, type-2 diabetes, cardiovascular disease and cancer. The findings of the study will make diagnosis easier and more efficient, reducing the risk of disease.

Researchers from Queen Mary University of London scanned the genomes of seven families experiencing delayed puberty. Their genetic profiles were analysed to identify specific genes that were different in these families, compared to individuals who started puberty normally. The researchers identified 15 candidate genes which were then examined in a further 288 individuals with late-onset puberty.

One gene was found to have common variants in nine families. The gene appears to contribute to the early development of gonadotropin-releasing hormone (GnRH) neurones in the brain. At puberty, a surge of GnRH is released, signalling to the pituitary gland to release further hormones that act on the ovaries and testes, triggering reproductive function (sexual maturation). If development of the GnRH neurones is delayed, the surge of GnRH that initiates these signals is also delayed.

Dr Sasha Howard who led the study said, “Studies estimate that 60-80% of variation in the timing of puberty is genetically determined, yet this is one of the first genes with major impact to be identified. This is an exciting finding as disturbed GnRH neuron development has never been linked to simple delayed puberty before, and may reveal a new biological pathway in the control of puberty timing.”

The group has also shown that the same gene may be responsible for completely blocking puberty - a condition known as hypogonadotropic hypogonadism. “We are now looking at how different mutations in the gene affect puberty in individuals, from delaying its onset to preventing it altogether, said Dr Howard. “By looking more closely at how such genes affect the timing of puberty, we will also learn more about gene-environment interactions, for example how endocrine-disrupting chemicals may contribute to early an onset of puberty,” she said.

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Notes for editors

1. For further information about the study please contact:

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2. The study *Novel genes influencing the timing of puberty* will be presented in poster format by Dr Sasha Howard *et al.* at the Society for Endocrinology's annual BES conference at 13:00 on Tuesday 25 March 2014.
3. For other press enquiries, please contact the Society for Endocrinology press office:

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4. The Society for Endocrinology's annual BES conference is held at the ACC Liverpool from 24 - 27 March 2014. BES features some of the world's leading basic and clinical endocrinologists who present their work. Journalists wishing to attend should contact Omar Jamshed at the Society for Endocrinology press office.
5. The Society for Endocrinology is a UK-based membership organisation representing a global community of scientists, clinicians and nurses who work with hormones. Together we aim to improve public health by advancing endocrine education and research, and engaging wider audiences with the science of hormones www.endocrinology.org