



PRESS RELEASE

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Danish researchers discover a genetic explanation of delayed puberty in girls

Minor variations in a specific gene explain why some girls enter puberty more than 7 months later than their friends. It is the first time researchers have shown that changes in one single gene can have such a huge impact on the timing of girls' puberty.

The study was led by physician Casper Hagen and Professor Anders Juul from the Department of Growth and Reproduction at Rigshospitalet. The researchers have examined 964 Copenhagen school girls. The girls have been clinically examined for signs of puberty and their blood samples were analysed for certain genetic variations.

The investigation will be published today in the internationally recognized magazine *Scientific Reports*, and tomorrow the researchers receive the prestigious European Henning Andersen Prize in Dublin for the research. The prize is awarded every year to the most significant research of the year within its field.

The underlying biology

The identified gene is involved in maturation of the eggs in the girl's ovaries. Professor Anders Juul explains:

"Girls with this genetic variation have eggs that tend to mature more slowly, even though the eggs are stimulated to the same extent as in other girls. Therefore, it takes longer time for the girls to produce the female sex hormones needed for breast development and for puberty to begin."

The age at which a girl enters puberty varies considerably. It is well known that a part of this variation has a genetic explanation. It is, however, the first time that researchers have been able to demonstrate, that one specific gene plays such a role for pubertal onset.

Early or late puberty

Delayed puberty is a relatively rare condition in girls. It is much more common that girls go into puberty at an early age.

"The aetiologies of early and late puberty are very different. With these new findings, we have identified a genetic cause for at least some cases of late puberty. We have, however, no genetic explanations for the many cases of early puberty that we see in the clinic. Also, the increase in incidence of early puberty suggests that lifestyle and environmental factors play an important role here, rather than genetics," says Professor Anders Juul

The diagnose of delayed puberty

If a girl is more than 13 years old before she enters puberty, she will be diagnosed with delayed puberty, and may receive hormonal therapy to induce pubertal maturation. This study also included a group of girls with this diagnosis. The results showed that whereas one in ten of the 964 Copenhagen girls had the specific genetic variation, the prevalence was one in four among girls diagnosed with delayed puberty.



Rigshospitalet

"We will definitely use this knowledge in the future when we examine girls with late puberty," says Professor Anders Juul

Late puberty is more frequent in boys than in girls. The next step will be to investigate if the same gene can explain late puberty in boys.

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