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Structural variants of chromosome 9: a possible association with hypogonadotropic hypogonadism.

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Abstract

We report two cases of structural variations of chromosome 9 associated with hypogonadotropic hypogonadism and azoospermia in adolescent boys. One patient also had a partially imperforated urethral meatus. Histological examination revealed that both had hypotrophic and underdeveloped testes. There was no LH and FSH response to LH-RH stimulation nor was there any response to naloxone tests. Basal and HCG stimulated plasma testosterone values were below normal prepubertal levels. As the administration of gonadotrophins did not improve the clinical and hormonal findings, alternative androgen therapy was necessary to achieve secondary sexual characteristics. Although they reached a good level of androgenization, their testes were still very small and azoospermia remained, as confirmed by repeated semen analyses. A possible association between chromosome 9 polymorphisms and hypothalamo-pituitary axis abnormalities is suggested. It is hypothesized that structural variants of chromosome 9 are not unrelated occurrences. Furthermore, and in view of the fact that they can lead to a high risk of azoospermia and infertility, such variants call for clinical investigation.

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