Should 45,X/46,XY boys with no or mild anomaly of external genitalia be investigated and followed up?

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Résumé en anglais	OBJECTIVE: Few studies of patients with a 45,X/46,XY mosaicism have considered those with normal male phenotype. The purpose of this study was to evaluate the clinical outcome of 45,X/46,XY boys born with normal or minor abnormalities of external genitalia, notably in terms of growth and pubertal development. METHODS: Retrospective longitudinal study of 40 patients followed between 1982 and 2017 in France. RESULTS: Twenty patients had a prenatal diagnosis, whereas 20 patients had a postnatal diagnosis, mainly for short stature. Most patients had stunted growth, with abnormal growth spurt during puberty and a mean adult height of 158 ± 7.6 cm, i.e. -2.3 DS with correction for target height. Seventy percent of patients presented Turner-like syndrome features including cardiac (6/23 patients investigated) and renal malformations (3/19 patients investigated). Twenty-two patients had minor abnormalities of external genitalia. One patient developed a testicular embryonic carcinoma, suggesting evidence of partial gonadal dysgenesis. Moreover, puberty occurred spontaneously in 93% of patients but 71% (= 5) of those evaluated at the end of puberty presented signs of declined Sertoli cell function (low inhibin B levels and increased FSH levels). CONCLUSION: This study emphasizes the need to identify and follow-up 45,X/46,XY patients born with normal male phenotype until adulthood, as they present similar prognosis than those born with severe genital anomalies. Currently, most patients are diagnosed in adulthood with azoospermia, consistent with our observations of decreased testicular function at the end of puberty. Early management of these patients may lead to fertility preservation strategies.
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