

Testicular pain as an initial complaint in patient with classic form of Klinefelter's syndrome: a case report

Identifier/Topic: EP-1315/ Clinical case reports - thyroid/others

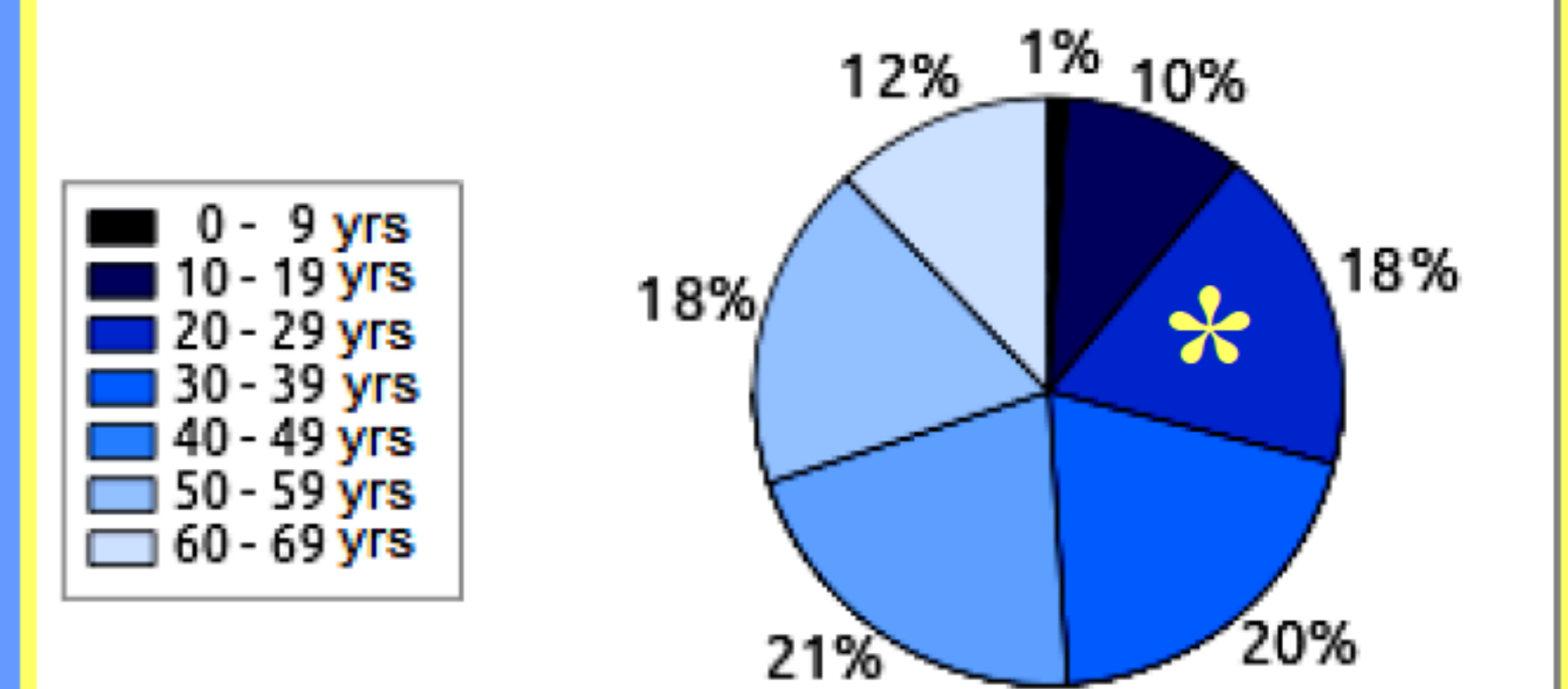
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Background:

In spite of being the most frequent genetic form of male hypogonadism, Klinefelter's syndrome is not so rarely undiagnosed until adulthood. As the severity of manifestations in Klinefelter's syndrome is proportional to the number of additional X chromosomes, they seem less severe and apparent in its classic form, with a 47,XXY karyotype than in cases of this syndrome with mosaicism and other variants.



Case report:

A 25-year-old male was referred for endocrinological evaluation by urologist because of the chronic testicular pain preventing effective testosterone replacement of already proven hypogonadism.

Physical examination: normal height, long limbs, muscular weakness, reduced or completely absent hair in the androgen-dependent regions, clinical signs of testicular dysgenesis.

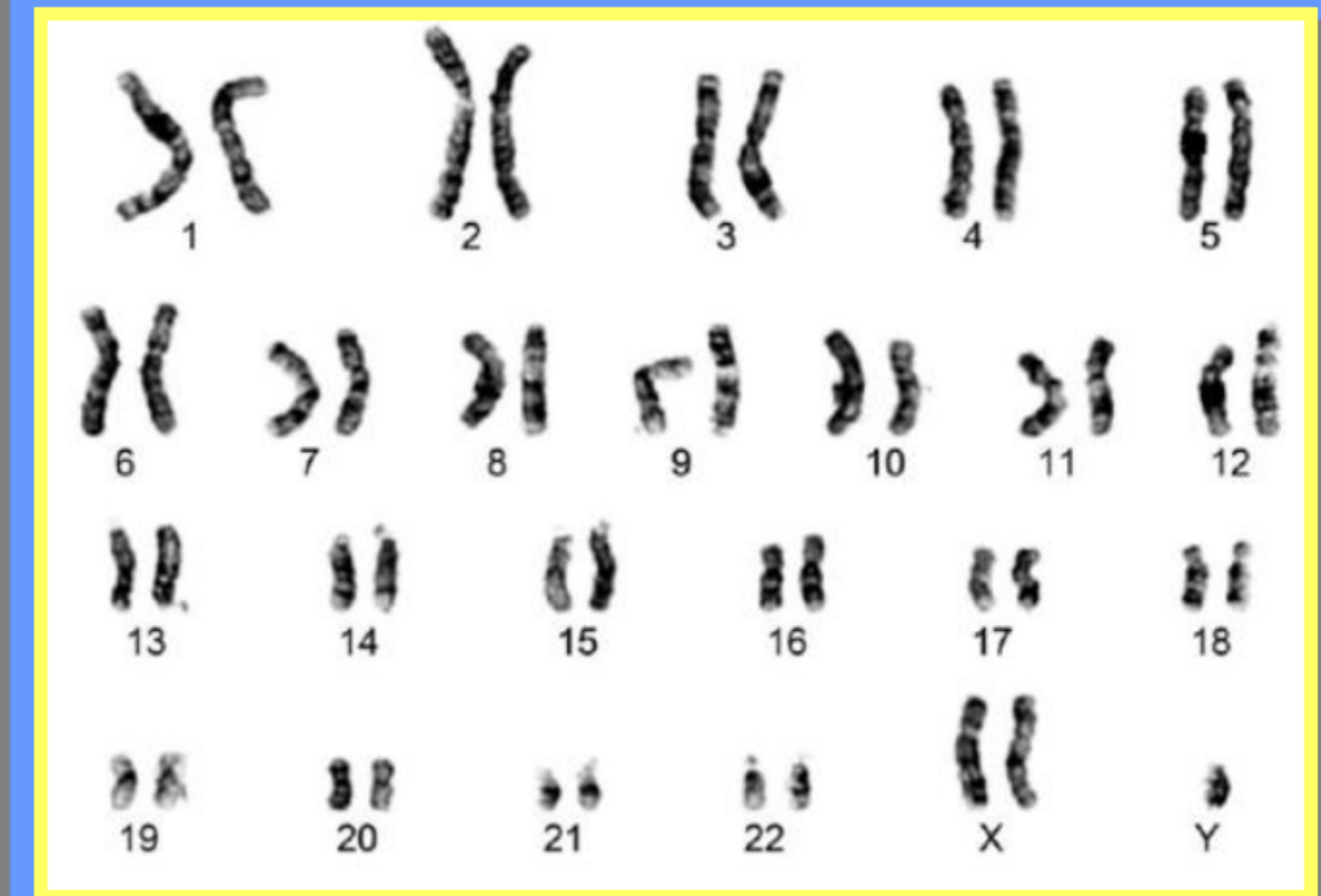
Hormone measurements: ACTH, 37.74 pg/ml; cortisol, 505.9 nmol/l; TSH, 1.81 μ IU/ml; fT4, 15.15 pmol/l; LH, 31.78 mIU/ml; FSH, 53.39 mIU/ml, free testosterone, 5.0 pg/ml; PRL, 519.9 mIU/l; vitamin D, 21.25 ng/ml.

Pituitary MRI: a profound arachnoid recessus pushing the pituitary toward the dorsum of the sella turcica;

Densitometry: osteopenia with the L1–L4 *T*-score of -2.7 and the hip *T*-score of -2.1 ;

Karyotype: the classic, 47,XXY form of Klinefelter's syndrome

Treatment and follow-up: testosterone replacement with the close follow-up of its effects.



Discussion:

Main issues for discussion:

- Universal screening vs case finding for earlier detection of disorder;
- Necessity of well-programmed care and multidisciplinary approach;
- Timing of testosterone treatment, treatment options and regular checkups interval and coverage;
- Fertility issues and a need for obligatory genetic testing.

Conclusions:

The case presented demonstrates a classical form of the Klinefelter's syndrome with not so classical presentation of testicular pain which halted the correction of proven androgen deficiency. Coexistent empty sella with hyperprolactinemia demand further periodical reassessment.

References:

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2. Nieschlag E: Klinefelter syndrome: the commonest form of hypogonadism, but often overlooked or untreated. *Dtsch Arztebl Int* 2013; 110(20): 347-53.
3. Groth KA, Skakkebaek A, Høst C, Gravholt CH, Bojesen A. Klinefelter Syndrome – A Clinical Update. *J Clin Endocrinol Metab* 2013; 98: 20-30.

