

A rare case of pseudoisodicentric X chromosome in a patient with primary amenorrhoea

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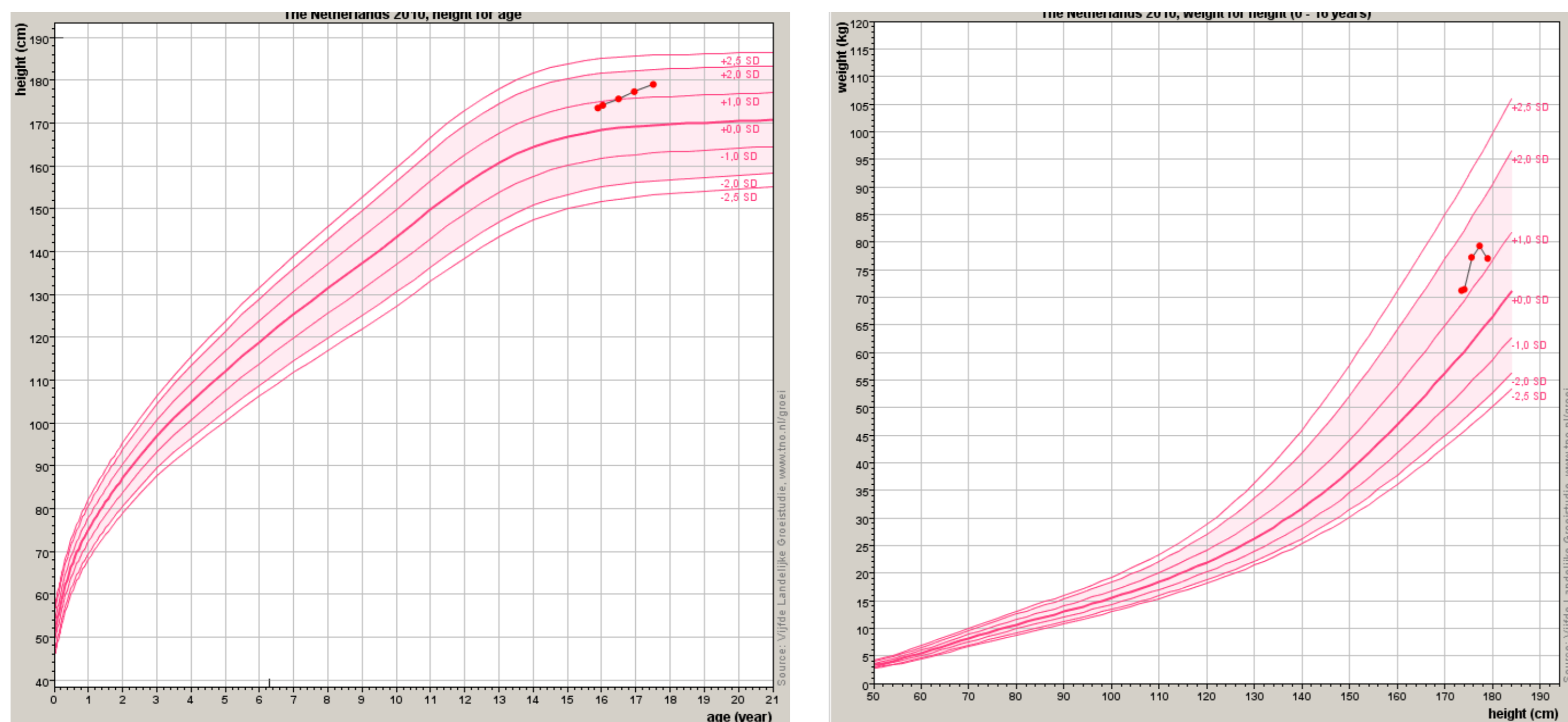
No potential conflicts of interest.

Background

Pseudoisodicentric X chromosomes with an Xq deletion (46,X,idic(Xq)) are rare. Most cases are mosaic, the other cell line being 45,X. Nonmosaicism is rare. Phenotype is characterized by the resultant of the X deletion. Variations from short to tall stature can occur and ovarian failure is a common feature.

Case description

A 16 year old girl was referred to our clinic with primary amenorrhoea. She was also known with learning disabilities. Physical examination showed a normal height (+0.8 SDS) and minimal pubertal development (A1, P2, M1-2). Laboratory testing demonstrated hypergonadotrophic hypogonadism.

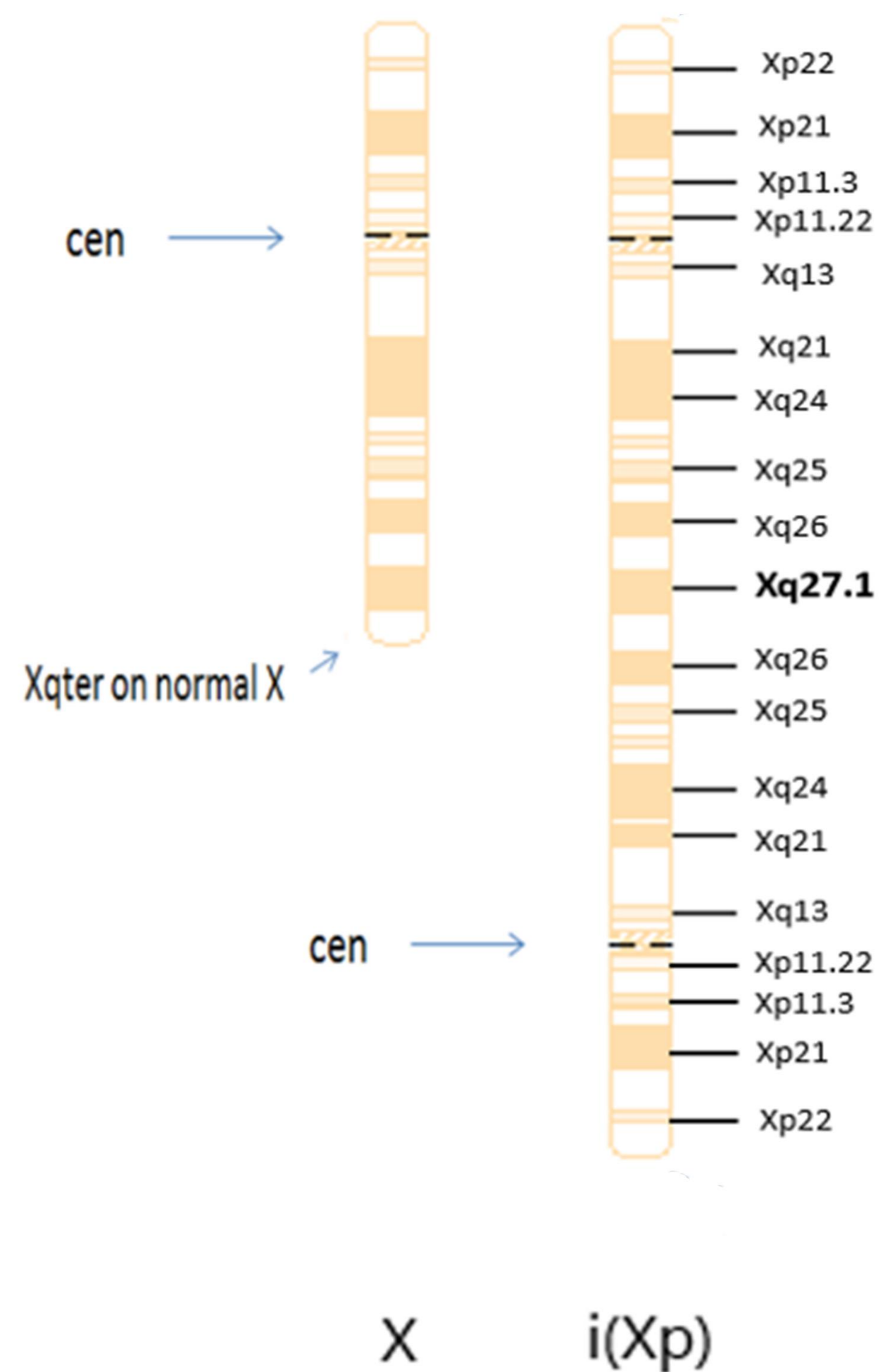


| Laboratory test | Results | Reference values |
|-----------------|---------------|--------------------|
| LH | 19,4 mU/mL | <15,8 mU/mL |
| FSH | 62,8 mU/mL | 0,8-8,5 mU/mL |
| Estradiol | < 0,06 nmol/L | 0,04 – 1,83 pmol/L |
| Progesterone | <0,6 nmol/L | 0,3 – 37,8 nmol/L |

Results

Conventional karyotyping of lymphocytes repeatedly revealed an aberrant X chromosome consisting of twice the short arm, twice a centromere and twice the long arm until Xq27, with a loss of a small part of the long arm of the X chromosome (nonmosaic 46,X,psu idic (X) (q27.1)). Genomic micro-array showed duplication of Xp22.33q27.1 and deletion of Xq27.2q28.

Genetic results



Conclusion

In our patient with nonmosaic 46,X,psu idic (X)(q27), the presence of only a small deletion of Xq in combination with a normal height, supports the 'inactivation theory' first proposed in 1994. Loss of genetic material of the X chromosome distal from q27.1 led to ovarian failure and did not inactivate SHOX, leading to normal height. However, the exact mechanism still remains to be discovered.



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