

A Case of Panhypopituitarism With SOX3 Gene Deletion

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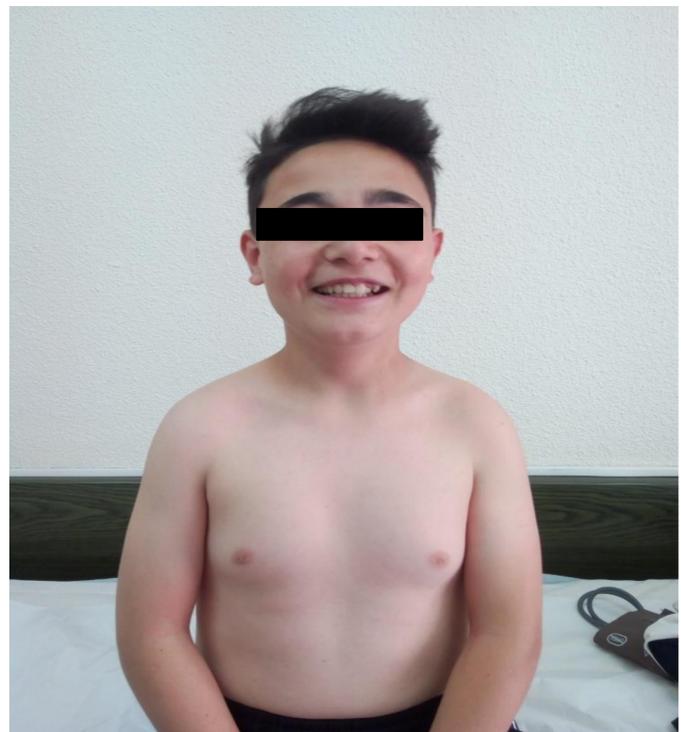
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Introduction:

It is known that microduplications including the SOX3 gene and intragenic duplications leading to loss of function in the gene cause panhypopituitarism, which can be accompanied by intellectual failure. Here, we report the first known case of panhypopituitarism, a deletion of the X chromosome, including the SOX3 gene in the q27.1q27.3 region.

Case

A 15-years and two months old male patient was referred to our clinic because of short stature, delay in puberty, and a reversal of the bone age. He had a history of mild mental retardation. He were born 3000 gr in term. There was no consanguinity between the parents. Physical examination revealed weight: 45,7 kg (<3p), height: 141,8 cm (<3p), height SDS -4.14, bone age 11y, 4-5. metacarpal shortening, low-narrow shoulder, scrotum hypoplasia, testicular volume was 6 / 6 ml. Karyotype analysis was 46, XY. IGF1: 60.4 mg / dl (<-3SD), IGFBP3: 3297 (-2, -3SD), L-Dopa test peak growth hormone 0.66 mg / dl, clonidine test peak growth hormone 0.98 mg / dl. growth hormone treatment was started due to hormone deficiency. In the LHRH test, peak FSH: 3.55 U / L, peak LH: 3.9 U / L was detected.



Findings

In the CGH analysis of the patient, there was a 3,961 kb deletion in the q27.1q27.3 region of the X chromosome containing the SOX3 gene.

Discussion

It is known that duplications containing SOX3 gene in the q27 band region of X chromosome cause panhypopituitarism, short stature and learning difficulties. In addition, intragenic duplications that lead to loss of function in the SOX3 gene cause panhypopituitarism. In other words, both high dosage and loss of function in the SOX3 gene are responsible for panhypopituitarism. In our patient, the deletion which caused null variance in SOX3 gene was thought to be responsible for panhypopituitarism, behavioral problems and speech deceleration. Our case is important in terms of being the first patient with SOX3 nulldelessness and panhypopituitarism.

