TWO CASES OF NON-SYNDROMIC CONGENITAL UNILATERAL BREAST HYPOPLASIA IN ONE FAMILY

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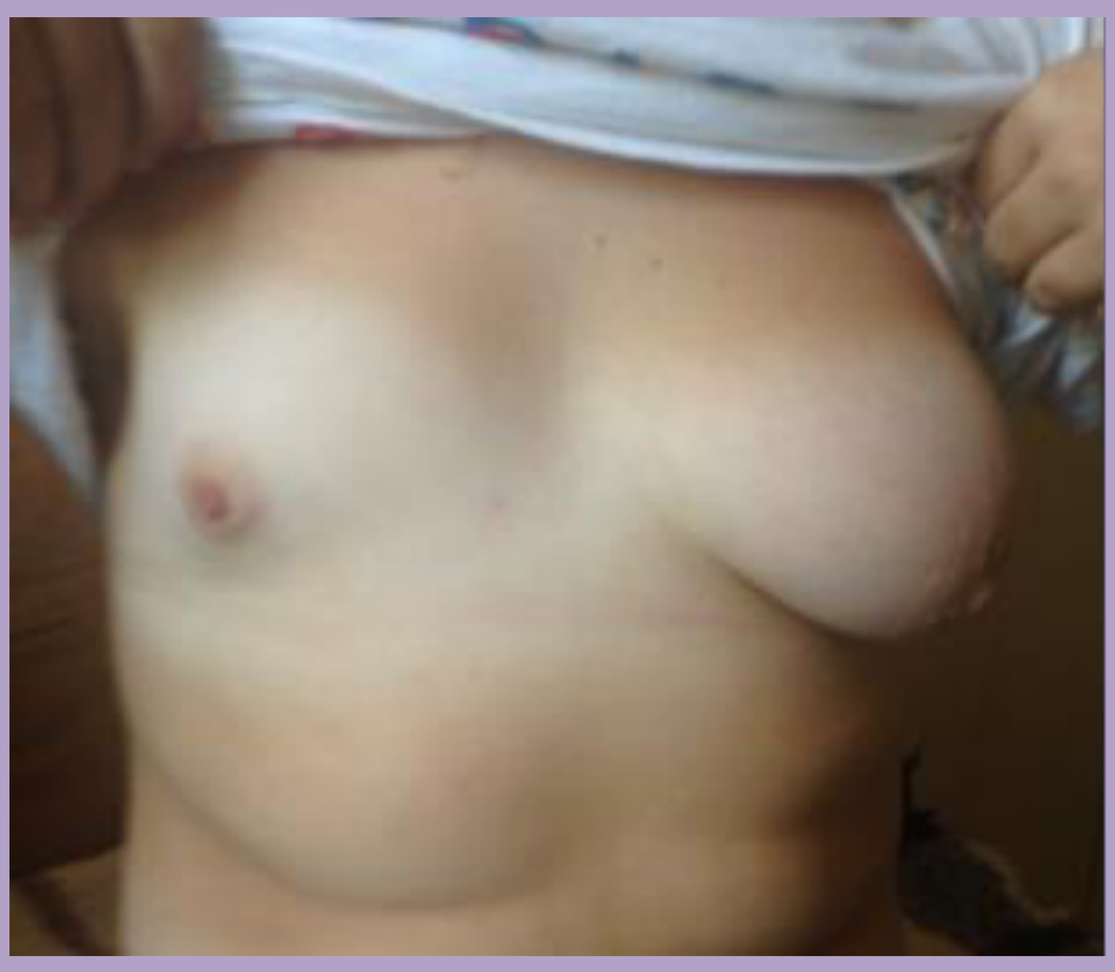
Introduction

Micromastia or breast hypoplasia is a condition which is described as postpubertal underdevelopment of a woman's mammary tissue. Breast hypoplasia may be congenital or acquired. The defect can be isolated or associated with other pathology, including syndromes and chest wall anomalies, it can also be unilateral or bilateral. Unilateral congenital breast hypoplasia is a rare anomaly of breast development, whose incidence is unclear. .



Results

The ultrasound of the breasts showed hypoplasia of the mammary tissue on the right breast while the other breast was normally developed (M5). Hormonal analyses showed normal estrogen and gonadotropin levels. The ultrasound of the gonads was uneventful and correspondent to her age. Also, there were no abnormalities of the chest wall. A whole exome sequencing was performed at the Genetics Department of the Technical University in Munich and it didn't show any mutations of the genes most commonly associated with this condition or any novel mutations. The most common syndromic causes for congenital breast hypoplasia, Poland's and Turner's syndrome were excluded.

Methods

We present a case of a 15 old girl referred to the Pediatric Endocrinology Department by the child's family doctor due to micromastia of the right breast. Pubertal Tanner stage was G3, she had her period at the age of 13 and the menstrual cycles were regular and normal. Her height was on the 50th percentile,while her weight was on the 75th percentile. The patient was otherwise healthy. From the family history her maternal grandmother had the same condition,which was never examined further or treated .

Conclusion

Isolated congenital unilateral micromastia is a very rare condition with unknown incidence. Few gene mutations have been implicated as the most common culprits causative of the the non-syndromic cases. Since no mutations were detected in our case we hypothesize that other mechanisms can be responsible for the condition. Some authors have suggested that congenital unilateral hypoplasia of the breast may be caused by under-expression of the estrogen receptors in the breast, but this theory needs further clinical evaluation . The treatment is surgical reconstruction of the affected breast after the child reaches a certain age.

References

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