Leydig Cell Hypoplasia in Three Siblings in the Same Family

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Introduction

- Leydig cell hypoplasia (LCH) is a rare disease and one of the causes of male disorder of sexual differentiation (DSD). Inactivating mutations in the luteinizing hormone/chorionic gonadotropin receptor (LHCGR) gene can produce LCH.
- In this poster, we present three siblings with LCH based on the clinical and laboratory finndings and the molecular diagnosis.

Cases

- A seven-year-old child was brought to our hospital for inguinal gonads.
- Physical examination was characterized by a predominantly female phenotype, a blind-ending vagina, and no Mullerian structures.
- Chromosomal analysis revealed 46 XX karyotype.
- SRY gene was normal.
- The sequence analysis of the LHCGR gene showed a homozygous mutation (p.A483D c.1448C>A).
- When other siblings were examined, they were found to have the similar physical findings and the same genetic abnormality.

Conclusion

- Our cases show LHCGR gene mutations have unique charcteristics from patients to patients.