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CORNELIA DE LANGE SYNDROME: A CASE REPORT

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INTRODUCTION

CORNELIA DE LANGE Syndrome (CdLS) is a rare syndrome, first described in 1933 by the Dutch pediatrician CORNELIA DE LANGE. This syndrome represents a multi-systemic disorder with physical, cognitive and behavioral characteristics. It is evident in the typical or classical form (craniofacial appearance, short stature and limbs deformities). However, not all individuals with CdLS present with the classic phenotype, as the clinical presentation can vary widely, from mild to severe forms.

METHOD

We report a case of CORNELIA DE LANGE syndrome discovered at the age of 11.5 years.

OBSERVATION

An 11 and a half years old female patient, from a non-consanguineous marriage, with no notable pathological history apart from a school failure consulted for a short stature at -2.8 standard deviations (SD). The clinical examination found a malformative syndrome and hirsutism with a clinical score of 10 (of which 02 criteria were cardinal) evoking a CdLS. The biologic and radiologic explorations excluded all chronic or endocrine pathologies that could explain this statural delay and hirsutism. The search for a sensitive disorder revealed hypoacusis and amblyopia. In view of these findings, the diagnosis of CdLS was made according to the recommendations of the latest consensus on the diagnosis and management of CdLS (without recourse to molecular biology). Growth hormone treatment was not indicated because of the good response to the GH stimulation test by GLUCAGEN/PROPRANOLOL and the absence of marketing authorization for CdLS in MOROCCO.

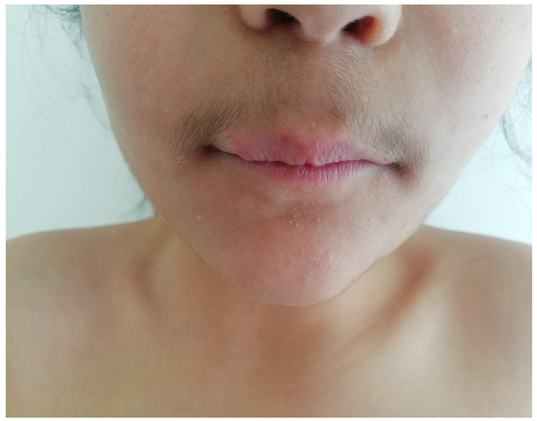

Image illustrating a long philtrum and downturned corners of mouth

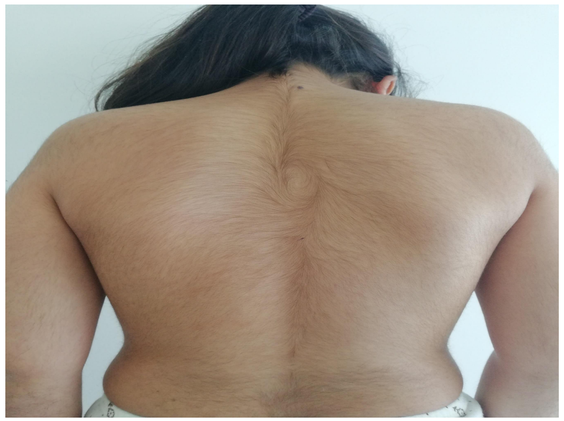

Image illustrating hirsutism

CONCLUSIONS

The recommendations provide a framework for improving the diagnosis and management of CdLS. CdLS is a complex disorder, in which many body systems are affected. It is important that a principal clinician be identified for each patient to ensure coordination of the many aspects of care in both childhood and adulthood. The proposed clinical and molecular diagnostic pathways are intended to be universally practiced.

REFERENCES

Kline, A.D., Moss, J.F., Selicorni, A. *et al.* Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. *Nat Rev Genet* **19,** 649–666 (2018).