# Novel phenotypic features of Loucks-Innes Syndrome, in one family from Saudi Arabia.

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## Introduction

Pathogenic variants in the DPH1 gene are causative of autosomal recessive developmental delay with short stature, dysmorphic features, and sparse hair syndrome) (DEDSSH) (OMIM 616901). Alternative title is Loucks-Innes syndrome.
8 cases reported in 2015, 4 cases by Alazami et al. (2015)from Saudi Arabia, and 4 cases by Loucks et al. (2015) from North America.
Our case series have more detailed phenotypic features not described before regarding Skeletal ,lung, and gastrointestinal anomalies.

## Objective

Outlining the phenotypic features of Loucks-Innes Syndrome as it has not been described fully in the literature..

## Methodology

Observational case series

## Result

This case series from one family in Saudi Arabia delineate the detailed phenotypic picture of this wide spectrum syndrome.
Three children from one family have sandal gap, single simian creases, normal nails,dolicocephaly,dysmorphic facial features in the form of upslanting eyes, hypertelorism, depressed nasal bridge, low set ears, sparse eyebrows, sparse eyelashes, bitemporal baldness that is progressive over time, and Post auricular skin tag.
All have mild congenital heart disease, and characteristic findings in the skeletal survey like non-ossified skull vault, small mandible, short neck, short nasal bone, non-ossified coccyx, mild bowing of proximal part of both ulna, ulnar deviation of wrists, hypoplastic radius with a dislocated bilateral elbow, and proximal radioulnar joints, short and broaden phalanges of toes, varus deformity of feet, multiple joint subluxations, accessory digits, bilateral acetabular dysplasia, and narrow chest with crowded 13 ribs.
Brain MRI had significant central nervous system (CNS) malformations like Blake pouch cyst, Dandy-Walker spectrum, and mild hypoplasia of splenium of the corpus callosum.
The patients have feeding intolerance due to severe GERD, swallowing difficulties, and repeated aspiration accompanied by the abnormal configuration of the duodenal C - loop with the ligament of Treitz seen at midline, and significant malrotation.
All of these patients have severe lung hypoplasia and the last patient who is 10 months old is oxygen-dependent since birth.
This syndrome causes severe failure to thrive and global developmental delay; the last patient cannot support the head, and no babbling at 10 months old.

Hearing assessment is normal ,but the patients have significant strabismus.
Karyotype and Microarray-Genomic Hybridization were normal for the three patients.
Whole-exome sequencing showed homozygous pathogenic variants in the DPH1 gene that support the diagnosis of autosomal-recessive developmental delay with short stature, dysmorphic facial features, and sparse hair syndrome.

## Conclusion

Figure1:detailed facial features of Loucks-Innes syndrome.

Loucks-Innes syndrome is characterized by multiple congenital anomalies that impact mainly the CNS, GIT, facial characteristics, cardiac, eye, hair,lung,skeletal, growth, development and most of the patient died in early childhood.