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EC PAEDIATRICS
Case Report

# A 14 Years Old Boy with Old Hands and Dysmorphism

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## Abstract

Microdeletion of chromosome 15q24 is a rare disorder with intellectual disability and clinical features, like coarse face, hands anomalies, ears malformation and sexual development delay.

We present a case of a 14 years old boy with coarse face, old hands, hypogonadism and learning problems.

The array test confirmed microdeletion of 15q24.

Genetic counselling and prenatal diagnosis are recommended for the next pregnancy.

***Keywords**: 15q24; Developmental Delay; Dysmorphism; Old Hands; Hypogonadism*

## Background

Microdeletion of chromosome 15q24 is a rare disorder with intellectual disability and clinical features, like coarse face, ears malformation and sexual development delay.

We present a case of a 14 years old boy with coarse face, old hands, hypogonadism and learning problems.

The array test confirmed microdeletion of 15q24.

This disorder also can have other presentation like seizures, cortical abnormalities, enlarged ventricles, thin corpus callosum, attention deficit, hyperactivity, autistic features, growth hormone deficiency and hypogonadotropic hypogonadism.

## Case Report

A 14 years old boy was observed in the pediatric consultation because he showed dysmorphic features, with coarse face associated with old hands for the age. He also have learning difficulties.

He is an only child of an unrelated young parents.

He borned by normal delivery with birth weight 3350g. Apgar score 10 at 5th minute.

No intercurrence during the neonatal period.

The hands was old for the age since first years of life.

On physical examination, the patient showed coarse face, long shape with epicanthal folds, downslanting palpebral fissures, high nasal bridge, smooth philtrum, and full lower lip. Eyes fundi - no changes.

Severe acne in the face.

Heart sounds are normal. Lungs sounds with no changes.

Abdominal region is normal too.

Testis 4 ml with microphalus.

Tanner stage: P2A1.

Bilateral old hands (Figure 1).

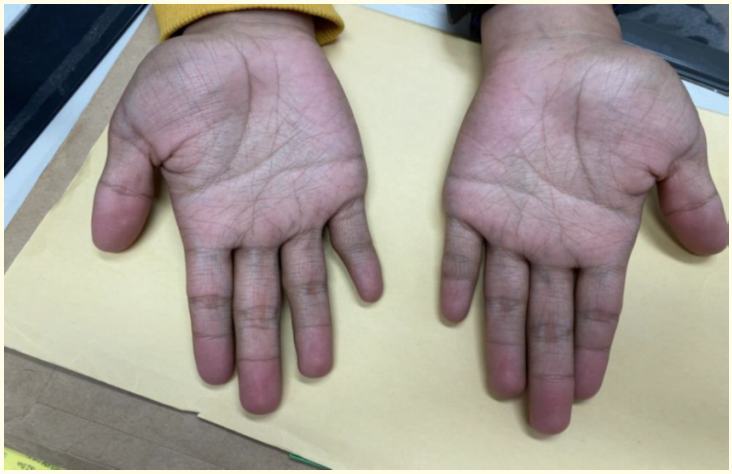

***Figure 1:*** *Old hands in a 14 years old boy with microdeletion of 15q24.*

Normal stature according to the mid-parental heigh. Heart/abdominal ultrasound was normal.

The rest of the examination was unremarkable.

Array test revealed microdeletion of 15q24.

Parents array test was normal too.

Heart/abdominal ultrasound - normal.

He started treatment with testosterone IM 100 mg every month with good response after 6 month: Tanner stage: P3A2 and testis 12 ml with increase lengh of penis size to normal percentile for the age.

## Discussion

When a patient showed development delay, we need to check the array test to exclude deletions or duplications of the chromosomes.

This is one of the indications for array test. The others are: mental retard, autism spectrum disorders and multiple congenital anomalies that do not characterize known genetic syndromes.

Array detected more 15% of chromosomes changes than a karyotype.

Our índex case, the patient have coarse face and this is important sign also in the storage diseases.

In this case the Gag and oligosacharides in the urine showed normal values.

The array test detected the deletion.

We can offer a prenatal diagnosis and genetic counseling for the couple in the future pregnancy.

Old hands are not a common presentation in any disease.

Hands with old signs associated with dysmorfic features are not common in del in 15q23.

According to the litetature, so far is not described [1-4].

## Conclusion

Whenever we find a patient with old hands without a reason, we need to exclude this disorder by doing a array that will or not confirm the diagnosis of microdeletion of 15q24.

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