

**POSTER PRESENTATION**

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# Case report of an adolescent girl with Kabuki syndrome and kyphoscoliosis, resistant at the conservative orthopedic treatment

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From 8th International Conference on Conservative Management of Spinal Deformities and SOSORT 2011 Annual Meeting  
Barcelona, Spain. 19-21 May 2011

## Background

The Kabuki syndrome (KS) is a rare genetic, hereditary, autosomic dominant, multiple anomaly syndrome, with an estimated incidence around 1-2/ 100 000 worldwide. Not all of the affected individuals have the same malformations.

Five major criteria delineate KS: postnatal short stature, skeletal anomalies, moderate mental retardation, dermatoglyphic anomalies, characteristic facial dysmorphism [1].

## Case report

We present the case of a 14 year girl diagnosed of the KS, referred to the Rehabilitation Service for kyphoscoliosis.

Searching in the literature, we didn't find reported the association with kyphosis, but only scoliosis and unspecific vertebral malformations.

The first referral to our service was on May 2010, when the patient is 14 years 10 months old. The clinical examination: occipito-sacral axis centred, plumbline of C7, T6, L4, S1 - 3/0/ 6.5/0 cm, Adam's Forward Bending test + (right rib cage elevated 2 cm), Hamstring tightness. Finger-floor distancy 25 cm.

The first Radiography: Hyperkyphosis 70°, Combined scoliosis T6-T12 12°, T11-L4 10°, Risser 2.

A first corse was prescribed - Maria Adelaida type, without correction neither of the scoliosis nor of the hyperkyphosis. A second corse Cheneau type was tried, without satisfactory results, and then a third corse, Michel Alegre modified type without improvement. The physiotherapy treatment was also prescribed. Presently,

the case is to be evaluated by Raquis Unity Rehabilitation and Orthopedic Surgery.

## Conclusions

Is the Kyphosis in our patient a characteristic of the Kabuki Syndrome? Is it a consequence of the Growth Hormone therapy our patient followed for 24 month for the short stature? Or is it a Scheuermann Disease accidentally associated with the Kabuki syndrome?

Published: 27 January 2012

## Reference

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doi:10.1186/1748-7161-7-S1-P4

**Cite this article as:** Taranu et al.: Case report of an adolescent girl with Kabuki syndrome and kyphoscoliosis, resistant at the conservative orthopedic treatment. *Scoliosis* 2012 **7**(Suppl 1):P4.

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