

## A case of bilateral dislocation of knees: Desbuquois dysplasia Kim variant

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### Introduction

Desbuquois syndrome (DBQD) is a rare form of osteochondrodysplasia (a disorder of the development of bones and cartilage)<sup>1</sup>. Variable degrees of short stature and short extremities, severe joint laxity and dislocation, osteopenia, kyphoscoliosis distinctive facial characteristics, are all possible<sup>2</sup>. Two forms have been distinguished based on the presence (type 1) or absence (type 2) of characteristic hand anomalies<sup>1</sup>. Short stature, joint abnormalities, characteristic facial features, and significant hand anomalies including short bones in the hands, long fingers, and advanced bone age are all hallmarks of the Kim variant of Desbuquois dysplasia, which has been described in 7 patients from Korea and Japan<sup>3</sup>. It is known that mutations in *CANT1* gene on chromosome 17q25 are responsible for both DBQD type 1 and the Kim variant<sup>1</sup>. While mutations in *XYLT1* are responsible for some cases of DBQD type 2<sup>1</sup>, the underlying cause in the majority of cases is still unknown. The condition follows an autosomal recessive pattern of inheritance<sup>3</sup>. Breathing difficulties of a severe nature have been linked to type 1<sup>4</sup>. Here, we report a case of Kim variant of Desbuquois dysplasia.

### Case report

A female baby was born via lower segment caesarean section (LSCS) to a primigravida mother of a consanguineous marriage. The baby, who was appropriate for gestational age, presented at birth with respiratory distress and bilateral hyper-extended knees. On examination, she had a round face, midface hypoplasia, short nose, flat nasal bridge, short neck and limbs, protuberant abdomen,

dislocation of both knees, and overriding of toes with flexion contractures at distal interphalangeal joints (Figures 1 and 2).



Figure 1: A newborn with characteristic facial features of Desbuquois dysplasia, short limbs, dislocation at bilateral knees, and overriding of toes with flexion contractures at distal interphalangeal joints



Figure 2: Overriding of toes with flexion contractures at distal interphalangeal joints

X-ray revealed generalised osteopenia, prominent greater and lesser trochanters, flat femoral heads, and short femoral necks producing a characteristic monkey wrench or Swedish key appearance (Figure 3), and dislocation of both knee joints (Figure 4). Short metacarpals, elongated phalanges, and remarkably advanced carpal bone age were also observed.

Molecular genetic testing by whole exome sequencing revealed a known missense variant c.467C>T p.(Ser156Phe) in *CANT1* gene in the homozygous state in the proband (ACMG classification- likely pathogenic). Sanger validation and parental segregation of the variant was not done. This variant is reported in HGMD. Biallelic variants in *CANT1* gene are known to be associated with Desbuquois dysplasia 1 (251450).

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**Figure 3:** X-ray showing generalized osteopenia, flat femoral head, prominent greater and lesser trochanters and short femoral necks producing characteristic monkey wrench or Swedish key appearance.



**Figure 4:** X-ray showing short limbs and dislocations of both knee joints

At present the child is under regular follow up in the Genetic and Orthopaedic clinics and is being monitored for growth parameters.

Less than 50 cases of Desbuquois dysplasia have been reported in the literature<sup>5</sup>. One should be aware of the risk factors and seriousness of this condition. Early detection and timely intervention of congenital dislocation of the knee could save the child from long-term disabilities and would help him/her to attain normal developmental milestones.

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