Sir,

Campomelic dysplasia (bent)-melia (limbs) is a rare (incidence 0.05–1.6 per 10,000), usually lethal congenital skeletal dysplasia with near total mortality in neonatal period.[1]

Our case was a 1.75 kg, small-for-gestational age (SGA) female child delivered to a third gravida mother by lower segment cesarean section (LSCS). Apgar scores at 1 and 5 min were 7 and 8 respectively. Antenatal USG at eight months of gestation was suggestive of skeletal dysplasia and oligohydroamnios. There was no history of consanguinity. Karyotype for female was normal, 46XX. Other two siblings were also normal. Further genetic studies were not done due to financial constraints.

On examination, the baby had respiratory distress, short birth length (45 cm), macrocephaly (head circumference 39 cm), micrognathia, low set ear, flat nasal bridge, hypertelorism, short and bowed lower limbs and pretibial skin dimpling on anterior side of tibia. Radiograph of the patient revealed bowed femur and tibia, hypoplastic scapula, absent parietal bone and large fontanelle [Figures 1 and 2]. USG of abdomen was normal. Septic screen was also negative. Baby expired on third day of life due to respiratory complications.

Campomelic dysplasia is diagnosed on the basis of clinical and radiological features.[2,3] Our patient fulfilled both clinical and radiological criteria.

Exact mode of inheritance is controversial (autosomal recessive/dominant), but the chromosomal abnormality in 17q resulting in abnormality in cartilage formation. In two-thirds of affected individuals with a 46, XY karyotype, male-to-female sex reversal had been described.[4] Most of the patients of campomelic dysplasia died in neonatal period due to severe respiratory distress which might be due to traceomalacia.[5] Campomelic dysplasia should be differentiated from thanatophoric dysplasia in which X-ray had the classical curved telephone receiver shaped femur.

As in the case of other neonatal lethal autosomal dominant disorders that have been thought to be autosomal recessive (e.g., osteogenesis imperfecta congenita), parents of infants with campomelic dysplasia had probably been often dissuaded from having further children as it results from new mutational event and has low risk of recurrence in subsequent pregnancies.
Letters to the Editor

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References


Sir,

An important problem in reproductive medicine is infertility. Many genetic disorders are mentioned as possible causes of human reproductive problem.\[1,2\] p53 codon 72 polymorphism is a genetic factor that is widely mentioned for its relationship with recurrent pregnancy loss and recurrent implantation failure.\[3,4\] However, the exact relationship is still controversial.

Here, the author performed a summative assessment and meta-analysis on the collected data to answer the question whether this polymorphism contribute to the problem of recurrent pregnancy loss and recurrent implantation failure or not.

This work is designed as a meta-analysis. The author performed a literature review to collect papers describing on p53 codon 72 polymorphism and its relationship with recurrent pregnancy loss and recurrent implantation failure. The searching was performed based on the standard referencing database, PubMed. The key words searched include "p53", "codon 72", "recurrent pregnancy loss" and "recurrent implantation failure". The papers with complete data in English were included for further meta-analysis.

A summative assessment on the number of subjects in each case and control group for each type of p53 codon 72 polymorphism was done. The correlation between p53 codon 72 polymorphism and the problem of reproduction, recurrent pregnancy loss and recurrent implantation failure was assessed by Chi-square test. P-value equal to or less than 0.05 is accepted as the statistical significant level in this work. In this study, a meta-analysis of two published reports addressing the same topic is performed.\[3,4\] There were 372 cases (302 recurrent pregnancy losses and 70 recurrent implantation failures) and 57 controls. There was no correlation between p53 codon 72 polymorphism and the studied problems of reproduction, recurrent pregnancy loss and recurrent implantation failure (\(P > 0.05\)).

It is no doubt that p53 is an important gene in medicine since it has proven relationships with cancer, longevity, reproduction, etc.\[5\] A genetic polymorphism, polymorphism of p53 codon 72, is widely studied for its role in reproductive medicine. However, the results on the correlation between polymorphism and abnormalities, recurrent pregnancy loss and recurrent implantation...