OSTEOGENESIS IMPERFECTA CONGENITA

(A Case Report)

by

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The incidence of the disease is quoted as 1:50,000. The basic error is a failure to form mature collagen resulting in derangement of energy metabolism coupled with immature cell function. This leads to biochemical disturbances as increased serum, urine and amniotic fluid in organic pyrophosphate levels, increased BMR and a decrease in platelet aggregation and sulphate uptake by skin fibroblasts. The resultant systemic change is failure of ossification and hence the pathological fractures.

In the event of a positive history of disease affecting one of the parents the chances of affected foetus are about fifty per cent. An affected pregnancy in a patient without any positive family history of the disease may pose interesting diagnostic and management problems. The present case illustrates them adequately.

Case Report

Patient K.D. aged 19 years, a primigrovida with history of 9 months of amenorrhoea was admitted in labour. Her general condition was good and all routine investigations were within normal limits. There was no history of pathological fractures or blue sclera in any of the members of the family as elicited later on. Abdominal examination revealed a full term, uniformly tense uterus through which no foetal parts could be identified. However, the foetal heart was heard at the left Mc Burny's point with a rate of 150 beats per minute. Vaginal examination revealed a long cervix admitting one finger, through which a soft pulpy part could be felt. Membranes were absent and pelvis was adequate.

A plain X-Ray abdomen was ordered to confirm the presentation but showed only a uniform soft tissue shadow in the lower abdomen corresponding with the uterus. No parts of foetal skeleton could be identified on the skiagram while the maternal skeleton was clearly outlined.

Labour was allowed to progress normally. The foetal heart suddenly disappeared after few hours. A still born male child weighing 4 lbs was delivered afer 14 hours of admission. Placenta and membranes were complete. The still born foetus (whose postmortom request was turned down by the relatives) had following features:

(1) globular soft head feeling like a bag of membranes without any palpable bone structure.

(2) Bulging eyes with bluish looking sclera.

(3) Flat, bowed and small (micromalia, extremeties.

(4 Body skin was thin and transluscent like a parchment membrane.

(5) Post mortom radiograph of the foetus (Figs. 1 and 2) showed:

(a) almost complete lack of calcification in the skull vault except a small streak like 'wormian' area of ossification.

(b) 'Waxy ribs' with thin cortex, poor calcification and multiple healed fractures looking like a 'string of beads'.

(c) Short and thick long bones with poor calcification and thin cortex associated with marked bending and distortion due to multiple fractures and callus formation.

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See Figs. on Art Paper III