

Prenatal ultrasonic findings of a child with congenital ichthyosis

A CASE Report

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Introduction

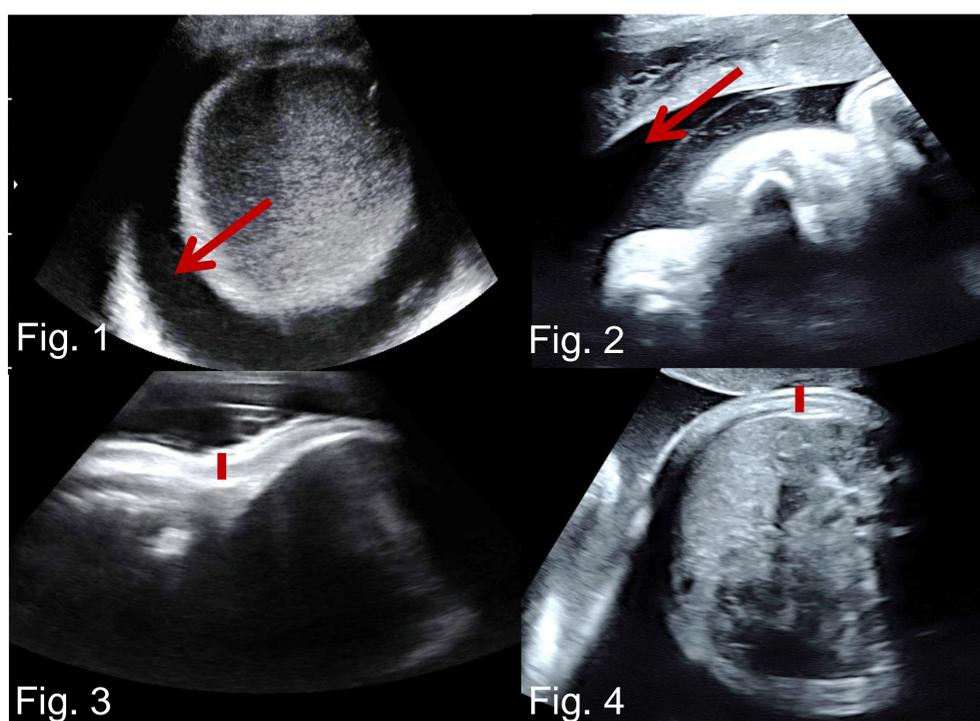
Congenital ichthyosis is a rare skin disorder of keratinization or cornification due to autosomal recessive or dominant mutation of certain genes (TGM 1, KRT 1, ALOXE 3, ABCA 12). The presence of dry surface scales is responsible for its naming as “collodion baby”. The probability for this disease is 1:300 000.

A CASE Report

A 41 year-old Gravida V, Para II came to Cantonal Hospital in St. Gallen for the first time in 37+6 pregnancy weeks. Her previous births were uncomplicated. She had a normal pregnancy and came with premature rupture of membranes and contractions. Surprisingly the performed ultrasound showed numerous abnormalities. Polyhydramnios was detected with an amnion fluid index of 22cm. Additionally the whole amniotic cavity except the placental site seemed to be detached from the myometrium (Figure 1 & 2). The amnion fluid surrounding all the foetal structures and umbilical cord showed excessive intra-amniotic debris.

Interestingly there was a hypo-echoic liquid layer between the myometrium and this area. Furthermore the foetal skin was thickened (Figure 3 & 4) with generalized oedema

In course of parturition a secondary caesarean section had to be performed because of stagnation during dilation phase. The primary cardiopulmonary adaptation of the new-born was normal. But the baby showed oedema all over her body especially on hands, feet, lips and genitalia. The skin was swollen and taut, very dry and parchment-like with fine scaling. The new-born could not close its eyes completely with bilateral ectropion and small eyelids. Furthermore the baby had everted lips, a reduced muscle tone in general and reduced joint movements (Figure 5). It was moved to the neonatal ward for closer observation and specific care with a high suspicion of congenital ichthyosis.



Discussion

There are sonographic markers for congenital ichthyosis including polyhydramnios, thickened skin and intra-amniotic debris