

Unusual findings in Down's syndrome: hand agenesis and hypospadias

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Objective

Heart, face and renal abnormalities are the most frequent Down syndrome abnormal features on ultrasound. Although rare, limb abnormality or hypospadias may also be found. Down syndrome must be discussed with the parents upon finding an increased NT with rare morphological features.

Methods

We report an unusual case of Down syndrome in a 38-year-old patient G2P0A1 with non-consanguineous marriage, and no relevant medical or family history. At 13 weeks, ultrasound confirmed an intrauterine viable pregnancy with fetal biometry of 12 weeks + 6 days of gestation, along with an increased nuchal translucency (4.9 mm) and a short fetal nasal bone. We suspected a hand agenesis since the hand was not seen clearly. Although an amniocentesis was proposed after discussing the case, the parents had refused any prenatal intervention and stated that regardless of the baby's condition, they will continue the pregnancy.

Results

Sonography at 17 weeks+ 6 days showed a male fetus with an adequate biometry. At this time, the diagnosis of hand agenesis was evident. There was also bilateral pyemic enlargement and the heart evaluation was also observed to be within normal limits. The situation was discussed once again with the parents who maintained their wish to continue the pregnancy but asked for amniocentesis. It showed a karyotype of a male fetus with Trisomy 21. Morphological ultrasound at 21 weeks confirmed prior findings about the hand agenesis and revealed the following additional features: atrioventricular canal, pelvic dilation, prefrontal thickening, and Sandal sign. The genitalia showed an abnormal appearance in favor of hypospadias. Following a spontaneous labor, a 3.1 kg baby was delivered at 39 weeks by cesarean for a pathological fetal heart rate with an Apgar score of 10 at one and five minutes. The baby was discharged at day 2 and is followed by a pediatrician. The patient's written consent was then obtained for scientific use of the data and images.

Conclusion

The association of hand agenesis and hypospadias in Down syndrome cases has not been reported in the medical literature. These unusual features should prove once again the extent to which actual Down syndrome cases can differ from textbook features.