Three cases of Fraser syndrome

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Objective

Fraser syndrome is also called as Cryptophthalmos-syndactyly syndrome or Cryptophthalmos syndrome. Fraser Syndrome is a rare and autosomal recessive(1). It is characterized by variable expression of cryptophthalmos, syndactyly, abnormal genitalia, malformations of the nose, ear and larynx, renal agenesis, clefting, skeletal defects (2,3,4). The aim of this case series was to study the consistent ultrasound findings and the post-abortal features of Fraser syndrome.

Methods

The cases with CHAOS (congenital high airway obstruction syndrome) and renal agenesis were followed with detailed postnatal examination of the abortus for syndactyly, ambiguous genitilia, cryptophthalmos and other major and minor criterias of Fraser syndrome. In the presence of these features, the genetic confirmation was done for mutation in FRAS, FREM 1 and FREM 2 gene.

Results

All the 3 cases had common antenatal finding of CHAOS, bilateral renal agenesis / dysplastic kidneys, small chin and ascites. Syndactyly, cryptophthalmos and ambiguous genitilia were not detected antenatally due to anhydramnios and gross ascites but were confirmed postnatally. In one women with non consanguinous marriage, there was a recurrence of Fraser syndrome.

Conclusion

We conclude that the cases with antenatal diagnosis of CHAOS and Bilateral renal agenesis/ dysplastic kidneys must be advised for proper postnatal examination of the fetus for the remaining features of fraser syndrome especially ambiguous genitilia, syndactyly and cryptophthalmos and then confirm genetically for proper counselling of the couple for future pregnancies.