Emanuel Syndrome with t (11;22) (q23;q11.2) Duplication and Diagnosed Prenatally: Case Report

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ABSTRACT

Introduction: Emanuel Syndrome is a rare chromosomal disorder caused by t der(11;22) (11q23;22q11), characterized by cleft palate, ear anomalies, heart defects, kidney abnormalities, genital anomalies, gastrointestinal anomalies, growth retardation, hypotonia and mental retardation.

Case Presentation: We present a case of Emanuel's Syndrome diagnosed prenatally with cleft palate, bifid uvula, long fingers, congenital diaphragmatic hernia and mega cisterna magna at 21 weeks of gestation.

34 years old, 21 weeks pregnant, gravida 3, parity 2, abortion 2. The patient was referred to the perinatology clinic with the diagnosis of congenital diaphragmatic hernia in obstetric ultrasonography. She had a history of bipolar disorder and asthma. There was no history of drug use in her current pregnancy. In the detailed perinatology examination of the patient, cleft palate, bifid uvula, long fingers, left diaphragmatic hernia, mega cisterna magna were observed. Amniocentesis was recommended to the patient. Amniocentesis result was reported as t(11;22) (q23;q11.2). Chromosome analysis was performed on the parents. It was found that the mother was a carrier of 47, X*, t der(22) t(11,22) (q23,q11). The father was found to have normal chromosome analysis. The patient was recommended to perform a feticide and then terminate. The pregnancy was terminated with medical treatment. Genetic consultation was requested for genetic counseling of the patient. The patient was discharged with recommendations.

Discussion: Emanuel Syndrome is a rare syndrome that can be diagnosed prenatally. In general, postnatal diagnosis can be made. The clinical findings of this syndrome occur due to 22q10-22q11 duplication and 11q23-qter duplication regions with excess t der(22). The unbalanced translocation occurs as a result of 3:1 meiotic chromosome missegregation during gametogenesis in the balanced translocation carrier. Balanced translocation carriers are clinically normal; these people are not suspected unless they have a child with t der(22) t (11,22) translocation, have had recurrent miscarriages or have infertility problems.

Conclusion: The extra chromosome is mostly passed on from mother or father to child. The surrogate parent has a normal chromosome number (46 chromosomes). However, there is an exchange of parts between one of the 11th chromosomes and one of the 22nd chromosomes. This is called a translocation. Because the gene is not gained or lost, it is called balanced translocation. That is, one of the parents is a balanced translocation carrier. If the parents are carriers, genetic counseling should be given. In order not to repeat it, prenatal genetic tests (prenatal tests) should be performed during pregnancy, and it is recommended to check the embryo before transfer with the Preimplantation Genetic Diagnosis (PGD) test. In addition, in the siblings of the carrier, care should be taken in terms of the syndrome and genetic examination should be performed.

Keywords: Emanuel Syndrome, prenatal diagnosis, Preimplantation Genetic Diagnosis

INTRODUCTION

Emanuel Syndrome is characterized by multiple congenital anomalies and growth restriction (1). It is caused by the presence of an additional genetic material of the chromosomes 11 and 22. There are very few publications in terms of clinical features, which causes us to have little information about the natural course of the disease.

Emanuel Syndrome is a rare chromosomal disorder caused by t der(11;22) (11q23;22q11), characterized by cleft palate, ear anomalies, heart defects, kidney abnormalities, genital anomalies, gastrointestinal anomalies, growth retardation, hypotonia and mental retardation. This syndrome can be diagnosed prenatally. We wanted to emphasize the importance of the Emanuel Syndrome, which was diagnosed prenatally with cleft palate, bifid uvula, long fingers, congenital diaphragmatic hernia and mega cisterna magna at 21 weeks of gestation.

CASE PRESENTATION

34 years old, 21 weeks pregnant, gravida 3, parity 2, abortion 2. The patient was referred to the perinatology clinic with the diagnosis of congenital diaphragmatic hernia in obstetric ultrasonography. She had a history of bipolar disorder and asthma. There was no history of drug use in her current pregnancy. In the detailed perinatology examination of the patient, cleft palate, bifid uvula, long fingers, left diaphragmatic hernia, mega cisterna magna were observed (Figure 1 and 2). Amniocentesis was recommended to the patient. Amniocentesis result was reported as t(11;22) (q23;q11.2). Chromosome analysis was performed on the parents. It was found that the mother was a carrier of 47, X*, t der(22) t(11,22)(q23,q11). The father was found to have normal chromosome analysis. The patient was recommended to perform a feticide and then terminate. The pregnancy was terminated with medical treatment (Figure 3). Genetic consultation was requested for genetic counseling of the patient. The patient was discharged with recommendations.

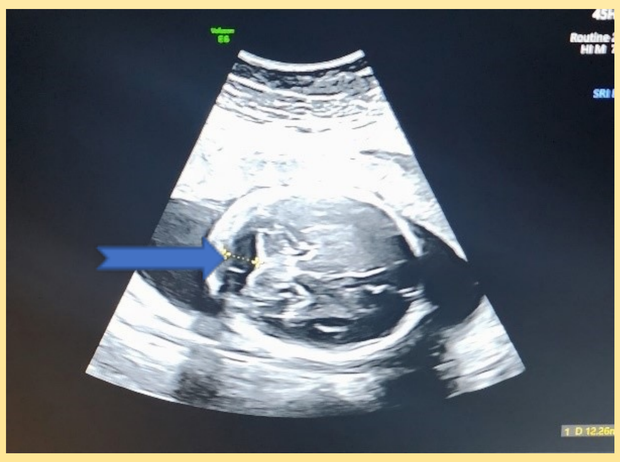

Figure 1: Blue arrow: pointing to mega cisterna magna

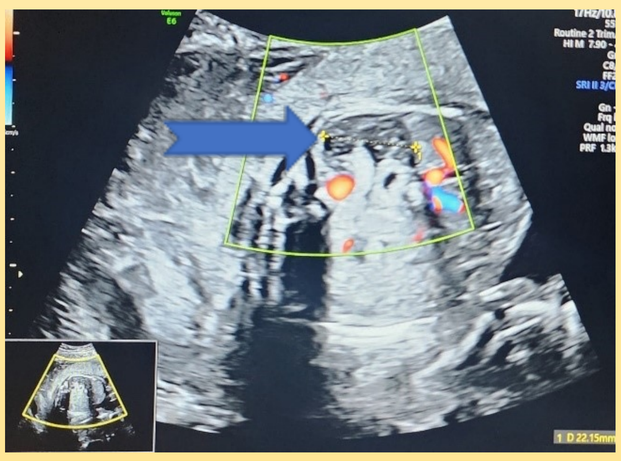

Figure 2: Blue arrow: left diaphragmatic hernia

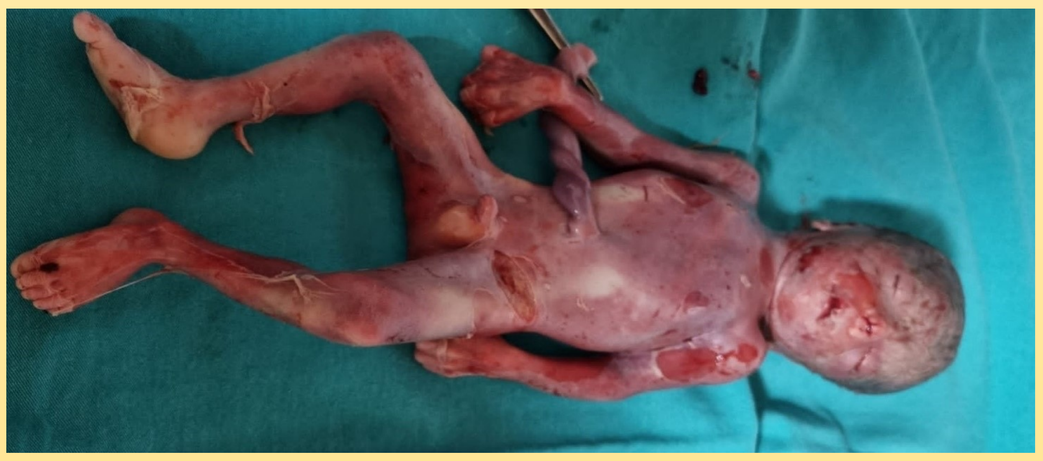

Figure 3: Postpartum view of the fetus

DISCUSSION

This genetic syndrome was named Emanuel Syndrome in 2004. The unbalanced translocation occurs as a result of 3:1 meiotic chromosome missegregation during gametogenesis in the balanced translocation carrier. Balanced translocation carriers are clinically normal; these people are not suspected unless they have a child with t der(22) t (11,22) translocation, have had recurrent miscarriages or have infertility problems. Carriers have up to a 10% chance of conceiving a child with this syndrome who survives to term (4). Heart defects, cleft palate, genitourinary tract malformations, intestinal atresia and craniofacial dysmorphism are well defined (5). Both antenatal and postnatal growth retardation occur in this syndrome. Although an exact mortality rate has not been reported, long-term survival is possible. The largest series on Emanuel Syndrome was published by Carter M et al. The families of 95% of these children had 11:22 translocations and, just like in our case, the mother of 90% was found to be a carrier. The most common complication during pregnancy was intrauterine growth retardation with 24%, followed by decrease in fetal movements with 18%, oligohydramnios with 16%, breech presentation with 14%, vaginal bleeding with 11% and prematurity with 9%. Ultrasound anomalies were detected in 16% of cases as heart, brain and renal malformations. This rate has been increasing in recent years due to the developments in ultrasonography.

CONCLUSION

The extra chromosome is mostly passed on from mother or father to child. The surrogate parent has a normal chromosome number (46 chromosomes). However, there is an exchange of parts between one of the 11th chromosomes and one of the 22nd chromosomes. This is called a translocation. Because the gene is not gained or lost, it is called balanced translocation. That is, one of the parents is a balanced translocation carrier. If the parents are carriers, genetic counseling should be given. In order not to repeat it, prenatal genetic tests (prenatal tests) should be performed during pregnancy, and it is recommended to check the embryo before transfer with the Preimplantation Genetic Diagnosis (PGD) test. In addition, in the siblings of the carrier, care should be taken in terms of the syndrome and genetic examination should be performed.

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