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Antenatal Diagnosis of Trisomy 13: A Case Report

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Abstract **Case Report**

Introduction: Trisomy 13, a severe chromosomal anomaly with an incidence of 1/5000–1/20,000 live births, is characterized by multiple congenital malformations, including cleft lip and palate, renal anomalies, and polydactyly. Antenatal ultra- sound facilitates early diagnosis, guiding management decisions. ***Case Presentation:*** A 38-year-old primigravida's fetus was diagnosed with tri-somy 13 at 13 weeks via trophoblast biopsy, presenting bilateral cleft lip and palate, right megaureter, bilateral hydronephrosis, hexadactyly, and suspected Dandy- Walker malformation. Medical termination was performed at 20 weeks + 5 days. Results: Comprehensive ultrasound and genetic evaluation confirmed the diagno- sis, supporting informed decision-making for termination due to poor prognosis. ***Conclusion:*** This case underscores the critical role of antenatal ultrasound in de-tecting trisomy 13 and the importance of multidisciplinary counseling for complex congenital anomalies.

Keywords: Trisomy 13, cleft lip and palate, megaureter, hexadactyly, antenatal ultrasound, medical termination.

1. INTRODUCTION

Trisomy 13, also known as Patau syndrome, is a rare chromosomal disorder caused by an extra copy of chromosome 13, with an incidence of approximately 1/5000–1/20,000 live births [1]. It is associated with severe congenital malformations, including cran-iofacial anomalies (cleft lip and palate), central nervous system defects (holoprosen- cephaly, Dandy-Walker malformation), limb abnormalities (polydactyly), and genitouri- nary anomalies (megaureter, hydronephrosis) [2]. The prognosis is poor, with most af- fected infants succumbing within the first year due to cardiorespiratory or neurological complications [3].

Antenatal ultrasound is the cornerstone of diagnosis, detecting structural anomalies as early as the first trimester [4]. Specific markers, such as increased nuchal translucency, maxillary gap, and absent "superimposed-line" sign for secondary palate cleft, enhance diagnostic accuracy [5]. Trophoblast biopsy or amniocentesis confirms the karyotype, guiding parental counseling and management decisions, including medical termination in severe cases [6]. This case report describes the antenatal diagnosis of trisomy 13 in a fetus with multiple anomalies, including bilateral cleft lip and palate, right megau- reter, bilateral hydronephrosis, hexadactyly, and suspected Dandy-Walker malformation, highlighting ultrasound findings and clinical management.

2. CASE PRESENTATION

A 38-year-old primigravida presented for routine antenatal care. She had no significant medical or surgical history, no consanguinity, and regular menstrual cycles since menarche at age 13. Her blood group was B positive, with no alloimmunization (RAI ¡0). This was her first pregnancy (G1P0A0), spontaneously conceived, with the last menstrual period on 16 December 2024, corresponding to a gestational age of 20 weeks + 5 days at presentation.

First-trimester ultrasound at 13 weeks + 3 days, performed by a specialist (Dr. Chaab`en), revealed multiple anomalies: dilatation of the right renal pelvis and ureter suggestive of megaureter, mild left pyelectasis (4 mm), and hexadactyly of the right hand (Figure 1). The crown-rump length was 66 mm, and nuchal translucency was 2.1 mm, within normal limits. Additional findings included a bilateral cleft lip and palate and suspected ver- mian agenesis, raising concern for Dandy-Walker malformation. Trophoblast biopsy at 13 weeks confirmed a 46, XY karyotype with trisomy 13.

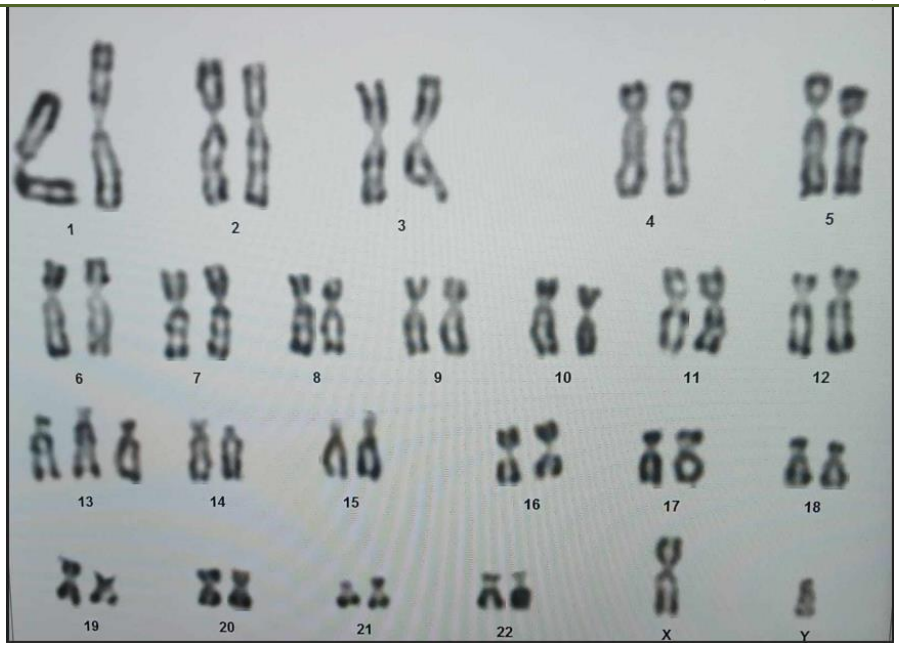

Figure 1: Genetic confirmation of Trisomy 13

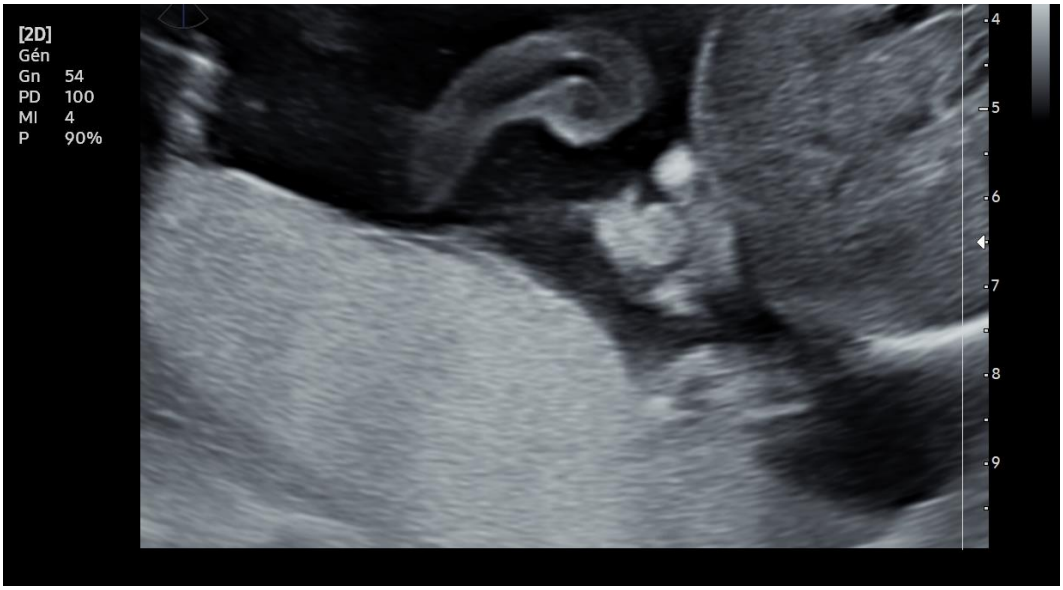

Figure 2: Transverse ultrasound at 20 weeks showing bilateral cleft lip and palate with disrupted upper lip continuity

Follow-up ultrasound at 20 weeks + 5 days confirmed the findings: bilateral cleft lip and palate with noncontinuous upper lip, right-sided grade 2 hydronephrosis with megaureter (ureteral dilatation not quantified), left pyelectasis (4 mm), and right hand hexadactyly. The ureterovesical junction was not visualized, supporting a diagnosis of congenital megaureter. The bladder was normal, and amniotic fluid volume was adequate (placenta normal, no polyhydramnios). Suspected vermian agenesis persisted, suggestive of Dandy-Walker malformation. Laboratory results showed hemoglobin of 14.6 g/dL, platelets of 189,000/µL, and gestational diabetes (fasting glu- cose 1.04 g/L, managed with diet).

Clinical examination revealed a stable maternal condition (GCS 15/15, temperature 37°C, blood pressure 120/70 mmHg, heart rate 88 bpm). The abdomen was soft, the uterus relaxed, and fetal movements were present. Vaginal examination showed a closed cervix with no bleeding or discharge. Given the severe prognosis of trisomy 13, including high risks of perinatal mortality and neurodevelopmental impairment, the patient opted for medical termination of pregnancy (IMG) after multidisciplinary counseling involving maternal-fetal medicine, genetics, and neonatology. The procedure was performed at 20 weeks + 5 days, and postmortem examination was declined.

3. DISCUSSION

Trisomy 13 is characterized by a wide spectrum of congenital anomalies, with 80–90 Cleft lip and palate result from failed fusion of the maxillary, nasal, and mandibular processes during the 4th–7th weeks of embryogenesis [8]. In this case, bilateral clefts were evident by 13 weeks, with disrupted lip continuity and palatal defects. Renal anomalies, including megaureter and hydronephrosis, are reported in 30–50

The suspected Dandy-Walker malformation, suggested by vermian agenesis, is less com- mon but reported in trisomy 13 [3]. However, diagnostic confirmation was limited by early termination, as MRI or postnatal imaging was not performed. Trophoblast biopsy at 13 weeks was pivotal, confirming trisomy 13 and guiding counseling [6]. The decision for medical termination reflects the poor prognosis, with a median survival of 7–10 days for live-born infants with trisomy 13 [3]. Ethical considerations in such cases balance parental autonomy, fetal prognosis, and psychosocial impacts, often favoring termination in severe cases [9].

Management of trisomy 13 involves multidisciplinary input, including geneticists, obste- tricians, and neonatologists, to provide accurate prognostic information [4]. Ultrasound surveillance monitors anomaly progression, while karyotyping via trophoblast biopsy or amniocentesis confirms the diagnosis. In this case, the absence of additional risk factors (e.g., consanguinity, maternal disease) and normal maternal hematological parameters (hemoglobin 14.6 g/dL) focused attention on fetal anomalies. Gestational diabetes, man- aged with diet, did not impact the outcome.

This case aligns with reported literature, such as a study by Chaoui *et al*., which high- lighted the maxillary gap and absent "superimposed-line" sign as early markers of cleft lip and palate [5]. The renal and limb findings are consistent with Jones *et al*.'s description of trisomy 13 phenotypes [2]. Future research should refine ultrasound markers for early detection and explore prenatal interventions for less severe cases, though most trisomy 13 cases remain incompatible with long-term survival [10].

4. CONCLUSION

This case report describes the antenatal diagnosis of trisomy 13 in a fetus with bilat- eral cleft lip and palate, right megaureter, bilateral hydronephrosis, hexadactyly, and suspected Dandy-Walker malformation, leading to medical termination at 20 weeks + 5 days. Antenatal ultrasound and trophoblast biopsy were critical for diagnosis, enabling informed counseling and decision-making. The case highlights the severe phenotype of trisomy 13, the utility of early ultrasound markers (e.g., maxillary gap, cleft palate signs), and the need for multidisciplinary management. Clinicians should prioritize comprehen- sive ultrasound evaluation and genetic testing in suspected aneuploidy to guide parental choices and optimize care.

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