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Genetic Insights from Chromosomal Microarray Analysis: The Predictive Role of Ultrasonography in High-Risk Pregnancies

Kromozomal Mikroarray Analizinden Genetik Bilgiler: Yüksek Riskli Gebeliklerde Ultrasonografinin Tahmin Rolü

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ABSTRACT

Aim: This study aimed to investigate the predictive value of prenatal ultrasonography (USG) findings in detecting chromosomal abnormalities identified through chromosomal microarray analysis (CMA) in high-risk pregnancies.

Material and Methods: A retrospective analysis was conducted on 122 singleton high-risk pregnancies undergoing CMA between 2021 and 2022. High-risk status was based on advanced maternal age, abnormal screening results, USG anomalies, or relevant family history. A scoring system was applied to USG findings, assigning one point for each fetal system with an anomaly. CMA was performed as a first-tier test via amniocentesis. Maternal demographics, USG scores, CMA results, and pregnancy outcomes were recorded and analyzed.

Results: Abnormal CMA results were detected in 34 (27.9%) cases. Maternal age was significantly higher in the abnormal CMA group (median 32 years vs. 29 years, p=0.030). Among pregnancies with abnormal CMA results, the rates of continuation and termination were equal at 44.1% (n=15). USG anomalies were significantly more prevalent in patients with abnormal CMA results, particularly in the abdominal (OR=2.84, 95% CI=1.01-7.96, p=0.041) and skin (OR=5.85, 95% CI=1.63-21.03, p=0.006) systems. Higher USG scores were significantly associated with abnormal CMA results (p<0.001). The most common chromosomal abnormalities were deletions (n=20, 58.8%) and duplications (n=10, 29.4%).

Conclusion: USG findings, especially system-specific anomalies and elevated USG scores, are significantly associated with abnormal CMA results. Advanced maternal age is also a predictive factor. Integrating USG scoring with CMA may enhance the diagnostic value in prenatal genetic assessment. Multicenter prospective studies are needed to validate these findings and improve clinical application.

Keywords: Chromosomal microarray analysis; prenatal diagnosis; high-risk pregnancy.

ÖZ

Amaç: Bu çalışmanın amacı, yüksek riskli gebeliklerde kromozomal mikrodizi analizi (CMA) ile saptanan kromozomal anomalilerin saptanmasında prenatal ultrasonografi (USG) bulgularının prediktif değerinin araştırılmasıdır.

Gereç ve Yöntemler: 2021 ve 2022 yılları arasında CMA yapılan 122 tekil yüksek riskli gebelik ile geriye dönük bir analiz yapıldı. Yüksek risk durumu; ileri anne yaşı, tarama testlerinde anormallikler, USG anomalileri veya aile öyküsüne dayanılarak tanımlandı. USG bulgularına, her bir fetal sistem anomalisi için bir puan olacak şekilde bir skorlama sistemi uygulandı. CMA, amniyosentezle birinci basamak test olarak yapıldı. Anne demografik verileri, USG skorları, CMA sonuçları ve gebelik sonuçları kaydedildi ve analiz edildi.

Bulgular: Anormal CMA sonuçları 34 (%27,9) olguda tespit edildi. Anormal CMA grubunda anne yaşı anlamlı olarak daha yüksekti (ortanca 32 yıl vs. 29 yıl, p=0,030). Anormal CMA sonuçları olan gebelikler arasında devam ve sonlandırma oranları %44,1 (n=15) ile eşitti. Anormal CMA sonuçlu hastalarda, özellikle abdominal (OR=2,84, %95 CI=1,01-7,96, p=0,041) ve cilt (OR=5,85, %95 CI=1,63-21,03, p=0,006) sistemlerinde olmak üzere, USG anomalileri anlamlı derecede daha yaygındı. Daha yüksek USG skorları anormal CMA sonuçlarıyla anlamlı bir derecede ilişkiliydi (p<0,001). En yaygın görülen kromozomal anomaliler delesyonlar (n=20, %58,8) ve duplikasyonlardı (n=10, %29,4).

Sonuç: USG bulguları, özellikle sisteme özgü anomaliler ve yüksek USG skorları, anormal CMA sonuçlarıyla anlamlı şekilde ilişkilidir. İleri anne yaşı da öngörücü bir faktördür. USG skorunun CMA ile birlikte değerlendirilmesi, prenatal genetik değerlendirmede tanı değerini artırabilir. Bu bulguları doğrulamak ve klinik uygulamayı iyileştirmek için çok merkezli prospektif çalışmalara ihtiyaç vardır.

Anahtar kelimeler: Kromozomal mikroarray analizi; prenatal tanı; yüksek riskli gebelik.

INTRODUCTION

Prenatal screening is essential to detect genetic and chromosomal anomalies early in pregnancy. In high-risk pregnancies or those with relevant risk factors, chromosomal microarray analysis (CMA) via amniocentesis is frequently used as a definitive diagnostic tool (1). Compared to traditional karyotyping, CMA provides higher resolution, allowing for identifying submicroscopic chromosomal abnormalities, such as microdeletions and microduplications (2).

The clinical utility of CMA in prenatal diagnosis remains a topic of debate in the literature. While some studies highlight its benefits in early diagnosis and prediction of postnatal developmental outcomes (3), others argue that its widespread use should be reconsidered due to increased costs and ethical considerations (4).

This study aimed to investigate the relation between CMA results and ultrasonography (USG) findings in high-risk pregnancies. USG is a fundamental tool for fetal anatomical assessment and plays a crucial role in detecting structural anomalies (5). We hypothesize that the likelihood of detecting genetic anomalies through CMA is higher in cases where USG findings indicate abnormalities. Accordingly, we will analyze the relationship between CMA results and USG findings to determine the predictive potential of ultrasonographic assessment in high-risk pregnancies.

MATERIAL AND METHODS

This retrospective study examined the results of amniocentesis and chorionic villus sampling (CVS) procedures performed between 2021 and 2022 at a tertiary referral center. In the study center, CMA was applied as a first-tier genetic test in high-risk pregnancies, without prior conventional karyotyping. Pregnancies included in the study were those identified as high-risk based on prenatal screening test results (e.g., increased risk for aneuploidy). Pregnancies were classified as high-risk if they met one or more of the following criteria: advanced maternal age (≥35 years), abnormal findings on first or second-trimester screening tests, structural anomalies detected via USG, or a positive family history for genetic disorders. In all these cases, amniocentesis was performed, and CMA was applied as a first-tier test without prior conventional karyotyping.

Fetuses from multiple pregnancies (n=2) and those with inconclusive CMA results (n=8), primarily due to sample contamination or technical errors requiring retesting, were excluded from the analysis. Thus, out of an initial cohort of 132 pregnancies, 10 were excluded, and data from 122 singleton pregnancies with definitive CMA results were included in the final analysis. Ultimately, 34 fetuses with abnormal CMA results and 80 with normal results were analyzed. The type of anomaly (e.g., deletion, duplication) was specified for fetuses with abnormal CMA results.

The demographic data (age), obstetric history (gravida, parity, number of abortions, number of living children), USG findings, CMA results, and pregnancy outcomes of the included patients were recorded.

Anomalies detected during the USG evaluation were categorized by system (central nervous system, cardiovascular system, genitourinary system, digestive system, extremities, etc.), with anomalies in each system defined as corresponding categorical variables. To quantify USG findings, a scoring system was developed. The USG scoring system used in this study assigned one point for each fetal system in which an anomaly was identified (e.g., cranial, cardiac, thoracic, skeletal, abdominal, genitourinary, umbilical/placental, or skin). The total score ranged from 0 to 7. This scoring system was developed specifically for the purposes of the current study and does not represent a previously validated or standardized metric. All USG assessments were performed and scored by a single experienced fetal medicine specialist blinded to the CMA results.

Ethical approval was obtained from the Ethics Committee of Toros University (Approval No: 2, Date: 24.01.2023).

Statistical Analysis

Data were analyzed using IBM SPSS Statistics version 22.0 (IBM Corp., Armonk, NY, USA). The normality of continuous variables was assessed using the Shapiro-Wilk test, and the homogeneity of variances was evaluated using Levene's test. For group comparisons, independent samples t-tests were used for normally distributed continuous variables, and the Mann-Whitney U test was applied to non-normally distributed ones. Categorical variables were compared using Pearson's chi-square test or Fisher's exact test, as determined by expected cell counts (Cochran's rule). For effect sizes, odds ratios (OR) with 95% confidence intervals (CI) were provided. All tests were two-sided with a type I error rate of 0.05.

RESULTS

Of the 122 pregnancies, 34 had abnormal and 80 had normal CMA results. The distribution of CMA results according to USG findings was summarized in a cross-tab to examine the association between sonographic anomalies and genetic outcomes (Table 1). Maternal age was significantly higher in pregnancies with abnormal CMA results compared to those with normal results (median age was 32 years vs. 29 years, respectively, p=0.030). No statistically significant differences were found between the two groups regarding gravida, parity, number of abortions, or number of living children (Table 2).

Table 1. Number of patients according to USG findings and CMA results

	USG Finding			Total (n=122)
	Normal (n=40)	Abnormal (n=80)	Other*** (n=2)	
CMA Result, n (%)				
Normal	29 (72.50)	49 (61.25)	2** (100)	80 (65.57)
Abnormal	8 (20.00)	26 (32.50)	0 (0.0)	34 (27.87)
Other	3* (7.50)	5* (6.25)	0 (0.0)	8 (6.56)

CMA: chromosomal microarray analysis, USG: ultrasonography, *: tests that require retesting or where contamination has been detected (excluded), **: multiple pregnancies (excluded), ***: chromosomal rearrangements, mosaic findings, and variants of uncertain significance that did not fit into deletion or duplication categories

Among 34 fetuses with abnormal CMA results, pregnancy continuation (delivery) and termination occurred at equal rates, each for 44.1% (n=15). Spontaneous abortion and unknown outcomes were observed equally in 5.9% (n=2) of cases each.

System-level analysis showed higher odds of anomalies in the abdominal (OR=2.84, 95% CI=1.01-7.96, p=0.041) and skin (OR=5.85, 95% CI=1.63-21.03, p=0.006) system USG findings among fetuses with abnormal CMA results. Differences in USG findings between normal and abnormal CMA results in other systems did not reach statistical significance (Table 3).

Among the 34 fetuses with abnormal CMA results, the deletion (n=20, 58.8%) was the most frequent chromosomal variation, followed by duplications (n=10, 29.4%), and other anomalies (n=4, 11.8%). A chi-square test showed a statistically significant deviation from equal distribution of anomaly types (p=0.004).

CMA results were compared according to USG scores. Abnormal array results were detected in 22.2% (n=8) of 36 fetuses with a score of 0, 23.9% (n=11) of 46 fetuses with a score of 1, 16.7% (n=2) of 12 fetuses with a score of 2, 62.5% (n=5) of 8 fetuses with a score of 3, and 66.7% (n=8) of 12 fetuses with a score of 4 (Table 4). When grouped as low (<3) and high (≥3) USG scores, the likelihood of abnormal CMA results was significantly higher in the high-score group (65.0% vs 22.3%, p<0.001). Further detail on the distribution of USG findings and pregnancy outcomes among fetuses with abnormal CMA results was provided in Table 5, supporting the observed correlation between specific sonographic patterns and genetic diagnoses.

Table 2. Distribution of obstetric history by CMA result

	Normal (n=80)	Abnormal (n=34)	p
Age (years)	29 (25-33) [17-42]	32 (28-38) [18-45]	**0.030**
Abortion	0 (0-0) [0-5]	0 (0-1) [0-3]	0.690
Gravida	2.5 (2-3) [1-7]	3 (2-4) [1-12]	0.730
Parity	1 (0-2) [0-4]	1 (0-2) [0-8]	0.290
Living	1 (0-2) [0-4]	1 (0-2) [0-8]	0.490

CMA: chromosomal microarray analysis

Table 3. System-specific distribution of USG anomalies by CMA result

	Normal (n=80)	Abnormal (n=34)	p
Cranial	25 (31.2)	14 (41.2)	0.420
Cardiac	19 (23.8)	12 (35.3)	0.300
Thorax	5 (6.2)	3 (8.8)	0.927
Placenta	1 (1.2)	0 (0.0)	1.000
Abdomen	9 (11.2)	9 (26.5)	**0.041**
Skin	4 (5.0)	8 (23.5)	**0.006**
Extremities	7 (8.8)	6 (17.6)	0.296
Umbilical cord	2 (2.5)	1 (2.9)	1.000
Fasciocranial	0 (0.0)	2 (5.9)	0.159
Vertebra	1 (1.2)	1 (2.9)	1.000
Urinary system	7 (8.8)	7 (20.6)	0.147

CMA: chromosomal microarray analysis, USG: ultrasonography

DISCUSSION

In this study, the relationship between CMA results obtained through amniocentesis and USG findings in pregnancies classified as high-risk by prenatal screening or clinical risk factors was evaluated. The results revealed that maternal age and USG scores were higher among pregnancies with abnormal CMA findings. In particular, anomalies in the cranial and cardiac systems were more prevalent in this group.

The observed association between increased maternal age and abnormal CMA outcomes aligns with prior studies indicating that advanced maternal age is a known risk factor for chromosomal abnormalities (1,6). Women aged 35 and above have a higher likelihood of fetuses affected by aneuploidies such as trisomy 21, 18, and 13 (7).

The findings of the present study are consistent with prior studies, underscoring the diagnostic relevance of USG anomalies in predicting genetic disorders. Studies by Fiorentino et al. (3) and Hsiao et al. (5) highlight how the systematic USG evaluation complements genetic testing, improving the overall diagnostic approach.

In addition to abdominal anomalies, cutaneous findings showed a strong association with abnormal CMA results. This is clinically relevant, as cutaneous anomalies such as increased nuchal translucency, cystic hygroma, and generalized skin edema are well-established ultrasonographic markers of chromosomal abnormalities. The present findings are in line with previous reports indicating that cutaneous markers, particularly in the first and second trimester, are predictive of pathogenic copy number variations and adverse genetic outcomes (3,8).

The significant association observed in the present study reinforces the importance of integrating cutaneous USG findings into prenatal risk stratification models when deciding on CMA testing.

In this study, ultrasonographic scoring was shown to play a relevant role in the stratification of genetic risk. The distribution of high USG scores was more common among fetuses with abnormal CMA results, supporting its predictive capacity.

The research by Cai et al. (8) highlighted the importance of USG scoring in identifying cases that benefit most from

Table 4. Distribution of abnormal CMA results according to USG score

	USG Score*					p
	0 (n=36)	**1 (n=46)**	**2 (n=12)**	**3 (n=8)**	**4 (n=12)**	
Abnormal CMA, n (%)	8 (22.2)	11 (23.9)	2 (16.7)	5 (62.5)	8 (66.7)	**<0.001**
	<3 (n=94)			**≥3 (n=20)**		
Abnormal CMA, n (%)	21 (22.3)			13 (65.0)		**<0.001**

CMA: chromosomal microarray analysis, USG: ultrasonography, *: each anatomical system with a USG anomaly was assigned 1 point, total USG scores were then categorized as <3 (scores of 0 to 2) and ≥3 (scores of 3 to 4) to evaluate their association with abnormal CMA results

Table 5. Genetic diagnoses and clinical features of fetuses with abnormal CMA results

CMA Results	n	Representative USG Findings	Outcomes
21q11.2q22.3 duplication (Down)	6	AVSD, Hydrops fetalis, Pericardial effusion, Echogenic lungs, Ascites, Hyperechoic bowel, Club foot, Cystic hygroma	5 Termination 1 Spontaneous abortion
18p11.32q23 duplication (Edwards)	4	Diaphragmatic hernia, Stomach in thorax, Heart displaced; Reverse a-wave in DV, Tricuspid leak, Pleural effusion, Spina bifida, Hydrops, Cystic hygroma, Ventriculomegaly, CP cyst, VSD, Multicystic kidney, Hypoplastic NB, Cystic hygroma	2 Termination 1 Delivery 1 Unknown
4p15.1 deletion	2	Inlet VSD, Omphalocele; Normal USG	1 Delivery 1 Termination
14q11.2 duplication (benign)	2	Increased CM; Ventriculomegaly	Termination
9q21.32 deletion	1	Increased NF, Hypertrophic small stomach, Pes equinovarus, hyperextended fingers, Strawberry-shaped skull	Termination
10q21.3 deletion	1	CP cyst, Echogenic intracardiac focus, Echogenic bowel, Dilated bladder, Dilated kidneys	Delivery
5p15.33p14.3 deletion + 20p13p12.2 duplication	1	Normal USG	Termination
6q16.3 deletion	1	Normal USG	Delivery
16p13.3 (Thalassemia trait)	1	Normal USG	Delivery
13q11q34 duplication (Patau syndrome)	1	Holoprosencephaly, Echogenic bowel, Hyperechoic kidneys	Termination
c.2187G>T in SOS1	1	Hypoplastic NB, Short and curved femur	Delivery
Xp22.33q28 duplication (Klinefelter)	1	Pelviectasia	Delivery
4q31.1 deletion	1	Pelviectasia	Delivery
Xp22.33q28 deletion (Turner)	1	Tricuspid regurgitation, Pleural effusion, Hydrops, NT: 7 mm	Termination
12p13.33p12.3 + 16p13.3 duplication	1	Ventriculomegaly, CC: 24 mm, Echoic bowel, SUA	Termination
(1-22)x2, (x)x1 (Turner)	1	Cystic hygroma	Unknown
4p15.1 + 12q21.1 deletion	1	Normal USG	Delivery
3p12.1 + 4p15.1 + 8q23.3 deletions	1	Normal USG	Delivery
Multiple deletions (4p15.1, 5q21.1, etc.)	1	Pericardial effusion, VSD, Cardiomegaly, Narrow thorax, Ascites, Rocker bottom foot	Spontaneous abortion
2p16.1 + 2q34 deletion	1	Normal USG	Delivery
12q21.31q21.32 + 13q31.2q31.3 deletion	1	Presence of a-wave in DV, NT: 5.4 mm, NB not observed	Delivery
3q26.1 + 12q21.1 deletion	1	Normal USG	Delivery
1p36.32 duplication	1	Intracardiac echogenic focus	Delivery
1q31.1q31.2 deletion	1	Pelviectasia	Delivery

CMA: chromosomal microarray analysis, USG: ultrasonography, AVSD: Atrioventricular septal defect, DV: Ductus venosus, CP: Choroid plexus, VSD: Ventricular septal defect, NB: Nasal bone, CM: Cisterna magna, NF: Nuchal fold, NT: Nuchal translucency, CC: Corpus callosum, SUA: single umbilical artery

CMA testing. Similarly, Fiorentino et al. (3) showed that system-based USG scoring improves the sensitivity and specificity of prenatal genetic screening.

Further support is provided by national studies, including Göktaş et al. (9), which demonstrate that organ-specific USG scoring correlates with increased likelihood of adverse genetic outcomes. Structured scoring may therefore improve the clinical utility of CMA by guiding its selective use in high-risk fetuses.

USG findings were associated with CMA results, supporting the utility of USG in prenatal genetic evaluation. Studies have shown that systematic ultrasonographic evaluation enhances the classification of genetic disorders (10). Scoring-based assessment of anomalies across organ systems provides a useful method for determining need for genetic analysis (3,8). The present findings support integrating system-based USG scoring into routine prenatal risk evaluation (10).

This study showed that decisions to continue or terminate pregnancies were evenly distributed among cases with abnormal CMA findings. This balance reflects not only the severity of diagnosed conditions but also the cultural, social, and psychological factors affecting parental decision-making.

Studies by Hammond et al. (4) and Hawkins et al. (11) have emphasized the critical role of counseling in navigating the ethical and emotional implications of prenatal diagnoses. The findings underline the need for interdisciplinary genetic counseling that includes obstetricians, geneticists, and psychologists to ensure families receive comprehensive, empathetic guidance.

Equal rates of continuation and termination in abnormal CMA cases reflect the clinical and ethical impact of prenatal genetic findings. Previous literature shows that detection of genetic abnormalities strongly influences decisions regarding pregnancy management (4,11). These results underscore the importance of comprehensive genetic counseling to support families in informed decision-making.

The clinical contribution of CMA extends beyond diagnosis. It informs decisions regarding prenatal care, delivery planning, and neonatal support.

In pregnancies with unclear sonographic findings or nonspecific anomalies, CMA may help clarify the underlying genetic etiology, thereby reducing uncertainty and guiding appropriate intervention strategies.

Although challenges remain in interpreting variants of uncertain significance (VUS), CMA remains an

indispensable tool in modern prenatal medicine, particularly when combined with detailed fetal imaging.

CMA allows for the detection of submicroscopic chromosomal abnormalities, offering greater resolution than conventional karyotyping (12,13). It contributes to early diagnosis of specific disorders and enables improved prediction of postnatal outcomes. This study supports the use of CMA in conjunction with USG to improve prenatal diagnostic accuracy. High USG scores (≥3), defined as anomalies in three or more anatomical systems, were found to be significantly associated with abnormal CMA results. This supports the potential role of multi-system USG involvement in predicting genetic abnormalities.

This study has several limitations. First, as a retrospective and single-center analysis, the findings may not be generalizable to broader populations. The lack of postnatal follow-up limits insights into the long-term developmental impact of detected anomalies. Additionally, the absence of standardized criteria for USG scoring introduces subjectivity into the evaluation process.

CONCLUSION

This study highlights the diagnostic value of prenatal ultrasonographic assessment in predicting chromosomal abnormalities detected via CMA in high-risk pregnancies. Elevated USG scores and the presence of specific system anomalies, particularly in the cranial and cardiac regions, were significantly associated with abnormal CMA results. Elevated USG scores, abdominal and cutaneous anomalies were significantly associated with abnormal CMA results. Advanced maternal age was also identified as an independent predictive factor. These findings emphasize the potential of integrating structured USG scoring with CMA to enhance early diagnosis, improve genetic counseling, and inform pregnancy management. Further prospective, multicenter studies are warranted to validate these results and standardize clinical protocols for prenatal genetic evaluation.

Ethics Committee Approval: The study was approved by the Scientific Research and Publication Ethics Committee of Toros University (24.01.2023, 2).

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