






Management and obstetric and perinatal outcomes of cystic hygromas

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ABSTRACT

Objective: Cystic hygroma is a congenital mono- or multiloculated cystic anomaly caused by abnormal fluid accumulation in the lymphatic system. It is the most common anomaly seen in the fetal back and neck, with an incidence of 1%. We aimed to determine the association between the diagnosis of cystic hygroma and adverse pregnancy outcomes.

Material and Methods: We retrospectively reviewed data on fetuses diagnosed with cystic hygroma by ultrasonography and evaluated demographic data, genetic results, anatomical screening results, and perinatal outcomes. In these cases, karyotyping was the only method available for genetic evaluation.

Results: Sixty pregnancies with a cystic hygroma diagnosis were included in the study. Fifty patients (83.3%) underwent invasive procedures, 25 fetuses (42.6%) had abnormal karyotypes, and the most common karyotype was trisomy 21. Twenty-six fetuses (43.3%) had congenital anomalies. Twenty fetuses were terminated, and 23 fetuses experienced intrauterine demise. Seventeen fetuses (28.3%) were delivered, and 5 (8.3%) were alive and without complications. Hydrops fetalis and structural anomalies were associated with worse outcomes, although statistical significance was limited by sample size.

Conclusion: Cystic hygroma coexists frequently with abnormal karyotypes and congenital anomalies. The fetal outcome is more favorable in the absence of additional adverse outcomes. Cytogenetic evaluation, fetal echocardiography, and second-trimester ultrasonography are essential for assessing prognosis and informing parents.

Keywords: Congenital anomalies, cystic hygroma, perinatal outcomes, prenatal diagnosis.

INTRODUCTION

Cystic hygroma is a mono- or multiloculated anomaly resulting from lymph accumulation in the jugular lymphatic sacs. It is predominantly located in the neck (75%), followed by the axilla (20%), retroperitoneum and intra-abdominal organs (2%), limbs and bones (2%), and mediastinum (1%).^[1] The overall incidence of cystic hygroma is approximately 1/1000-6000 in births and 1/750 in spontaneous abortions.^[2,3] On ultrasound examination, a cystic hygroma appears as a single or multilocular fluid-filled structure in the nuchal region or extending along the fetal body (Fig. 1).

Cystic hygromas have been associated with a wide spectrum of chromosomal abnormalities, including Turner syndrome, trisomy 21, trisomy 18, and trisomy 13, as well as various structural malformations.^[4-7] As the thickness of nuchal translucency increases, the likelihood of abnormal outcomes also rises.^[8] Although spontaneous regression may occur in some cases, most are associated with adverse outcomes such as fetal hydrops, intrauterine fetal demise, or termination of pregnancy.^[9,10] Given the variable clinical outcomes and their strong association with genetic and structural abnormalities, cystic hygroma remains a diagnostic and counseling challenge in perinatal medicine. In this study, we sought to report our institutional experience concerning the obstetric and perinatal follow-up and outcomes of cystic hygroma cases.

MATERIAL AND METHODS

This study retrospectively reviewed pregnancies diagnosed with cystic hygroma at Sakarya University Training and Research Hospital Perinatology Clinic between 2015 and 2020, approximately five years. Noninvasive ethical approval for this retrospective study was obtained from the Clinical Research Ethics Committee (Approval date: September 2020, number: 2020/500). All procedures followed the principles outlined in the Helsinki Declaration 2008. Data were collected by reviewing hospital records and contacting patients. Demographic characteristics of the patients, gestational age at diagnosis, invasive procedures performed, fetal karyotype results, congenital anomalies, pregnancy outcomes, and neonatal

outcomes were recorded. Ultrasound examinations were conducted using a GE Voluson E6 machine with the C4-8 curvilinear probe. Patients diagnosed with cystic hygroma received genetic counseling and were offered chorionic villus sampling (CVS) or amniocentesis based on gestational week. For CVS, 10-15 micrograms of placental tissue were obtained using a 20-gauge spinal needle and transported in a suitable medium. For amniocentesis, the initial 2 cc of amniotic fluid were discarded to reduce the risk of maternal contamination. Fluid was then aspirated into two separate syringes, 1 cc per week of gestation. Samples were analyzed by the Department of Medical Genetics using conventional karyotyping. Pregnancies that were ongoing underwent second-trimester ultrasonography and fetal echocardiography.

Statistical Analysis

Descriptive statistics were utilized. Comparative analyses included the Independent T-test, Mann-Whitney U test, Chi-square test, and binary logistic regression. The variables assessed in the statistical analyses included maternal age, gestational age at diagnosis (categorized as ≤ 14 weeks vs. >14 weeks), presence of hydrops, presence of congenital anomalies, and karyotype results (normal vs abnormal). We compared aneuploidy rates, pregnancy outcomes (termination, intrauterine demise, live birth), and neonatal outcomes according to these parameters. A p-value <0.05 was considered statistically significant.

RESULTS

Between 2015 and 2020, 62 patients were diagnosed with cystic hygroma over a five-year period. Two patients were excluded from the analysis due to incomplete data. The flowchart of patient inclusion, exclusion, and pregnancy outcomes is shown in Figure 2. The mean maternal age at diagnosis was 28.1 years (range: 18-41), and the mean gestational age at diagnosis was 13.7 weeks (range: 10-22). Invasive diagnostic procedures were performed in 50 patients (83.3%), while 10 patients declined karyotype analysis. Chorionic villus sampling (CVS) and amniocentesis were performed in 32 (64%) and 12 (24%) patients, respectively. Additionally, in

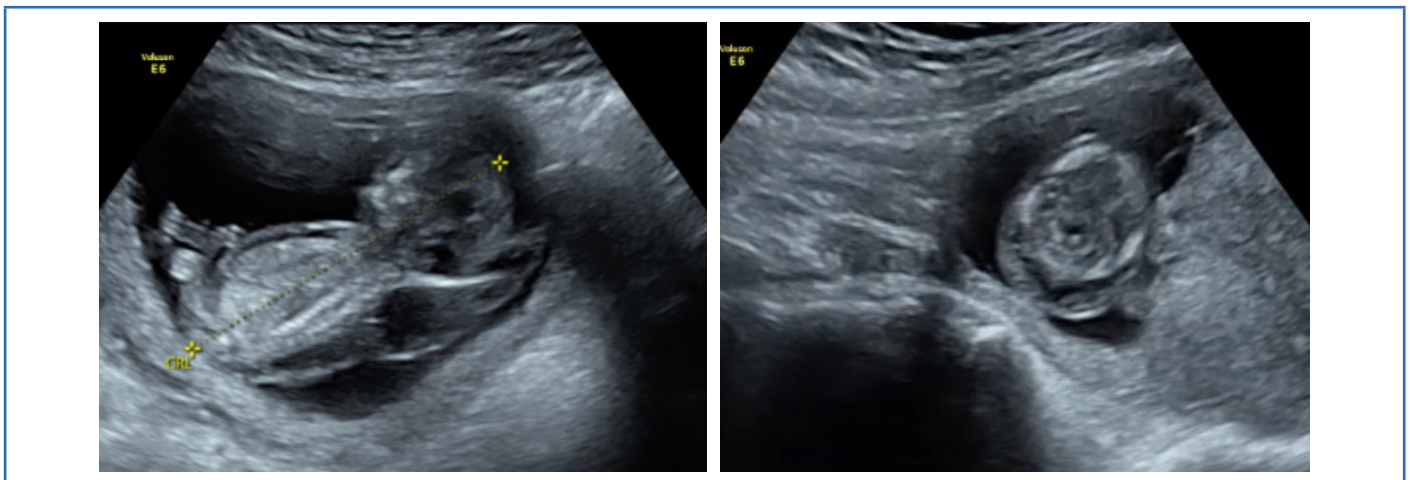


Figure 1: Sagittal and axial images of cystic hygroma.

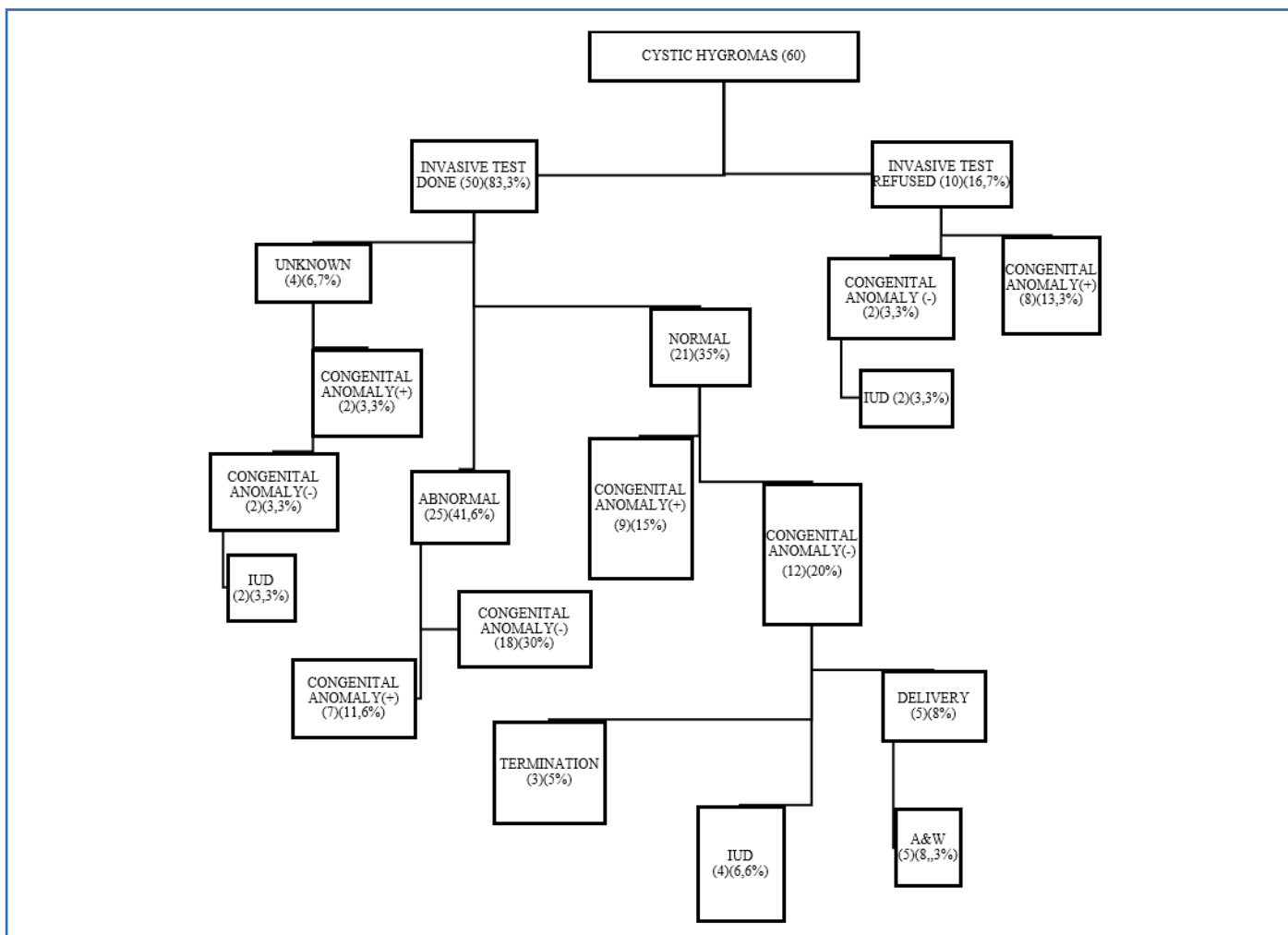


Figure 2: Flowchart of fetuses with cystic hygromas.

six patients (12%) who underwent CVS, amniocentesis was also performed because maternal contamination was suspected. No procedure-related complications were reported. In four cases, no karyotype results were obtained due to insufficient cell growth. Among the remaining patients, 25 (50%) were diagnosed with chromosomal abnormalities: trisomy 21 in 11 cases (22%), monosomy X in 10 cases (20%), and trisomy 18 in 4 cases (8%). Patients with abnormal karyotypes were significantly older than those with normal results (mean age: 29.5±7.1 vs. 25.7±4.4 years, p=0.035). Gestational age at diagnosis did not differ significantly between the two groups (13±2 vs. 14±2 weeks, p=0.635). Among patients diagnosed before 14 weeks, the prevalence of trisomy 21 and monosomy X was 20.6% (n=7) and 23.5% (n=8), respectively. In those diagnosed after 14 weeks, the rates were 33.3% (n=4) and 16.7% (n=2) (Table 1). Congenital anomalies were observed in 43.3% of the patients, with no significant difference between those with abnormal and normal karyotypes (42.9% and 28%, respectively, p=0.292) (Table 2). The most frequently observed anomalies were abdominal wall defects (19%), followed by limb anomalies (15.3%). Multiple anomalies were noted in 38.4% of affected fetuses. When stratified by gestational age at diagnosis (≤14 vs. >14 weeks), congenital anomaly rates

did not significantly differ (45.2% vs. 38.9%, p=0.649) (Table 2). Pregnancy outcomes included termination in 20 cases (33.3%), intrauterine fetal demise in 23 cases (38.3%), and live birth in 17 cases (28.3%). Among the live births, two neonates had monosomy X, four had trisomy 21, and the remainder had structural anomalies, including encephalocele, hydrocephalus, atrioventricular septal defects, cerebellar hypoplasia, and limb anomalies. Five neonates (8.3%) were delivered without perinatal complications and were classified as alive and well (Table 3).

Binary logistic regression was performed to identify predictors of live birth. Independent variables included karyotype status, presence of hydrops, gestational age at diagnosis (≤14 weeks vs. >14 weeks), and presence of congenital anomalies. Karyotype status was not significantly associated with live birth (OR=1.087, 95% CI: 0.194–6.101, p=0.924). The presence of hydrops was associated with a non-significant but substantial reduction in the likelihood of live birth (OR=0.126, 95% CI: 0.014–1.120, p=0.063). Congenital anomalies also trended toward reduced odds of live birth (OR=0.590, 95% CI: 0.165–2.118, p=0.419). Earlier diagnosis (≤14 weeks) showed a trend toward increased likelihood of live birth, though not statistically significant (OR=3.017, 95% CI: 0.686–13.259, p=0.144) (Table 4).

Table 1: Comparison of maternal age and gestational week at diagnosis according to normal and abnormal karyotype results

Karyotype	p	Maternal Age		Gestational Week	
			p		p
		Mean ±SD		Mean±SD	
Abnormal	25	29.52±7.125	0.035*	13±2	0.635**
Normal	21	25.71±4.417		14±2	

*independent t test p<0.05**Mann-Whitney U Test p value >0.05.

Table 2: Congenital anomaly frequencies according to karyotypes and gestational weeks at diagnosis

		Congenital Anomaly		p*
		No	Yes	
Karyotype	Abnormal	n	18	0.292
		%	72.0	
	Normal	n	12	
		%	57.1	
Gestational Week	≥14	n	23	0.649
		%	54.8	
	<14	n	11	
		%	61.1	

*Chi-square Test p value >0.05.

Table 4: Predictors of live birth in fetuses diagnosed with cystic hygroma

	OR	95% CI	p
KARYOTYPE (Normal vs. Abnormal)	1.087	0.194-6.101	0.924
Presence of hydrops	0.126	0.014-1.120	0.063
Presence of congenital anomalies	0.590	0.165-2.118	0.419
Gestational age at diagnosis (≤14 Weeks vs. >14 Weeks)	3.017	0.686-13.259	0.144

DISCUSSION

Cystic hygroma is associated with a high rate of chromosomal and structural abnormalities, leading to poor pregnancy outcomes. This study assessed the outcomes of pregnancies diagnosed with cystic hygroma. Consistent with existing literature,^[11,12] increased maternal age was associated with a higher prevalence of chromosomal abnormalities. Our findings align with those of Schreurs et al.,^[13] who also reported a low rate of normal karyotypes in fetuses diagnosed with cystic hygroma. Invasive procedures should be offered in all pregnancies with a cystic hygroma diagnosis because of the presence of abnormal karyotypes. Some patients refuse invasive diagnostic procedures due to their religious beliefs and fear of complications from the procedure. In our study group, 16.6% of the patients refused invasive procedures. An abnormal karyotype was present in 50% of the cases that underwent an invasive procedure, which is consistent with previous studies in the literature, reporting a range of 29% to 60%.^[11,14,15] In our cohort, 22% of abnormal karyotypes were trisomy 21, 20% were monosomy X, and 8% were trisomy 18; trisomy 21 was the most commonly observed aneuploidy. In contrast, some studies in the literature reported monosomy X as the most prevalent,^[13,14] followed by trisomy

Table 3: The outcome of cystic hygroma according to congenital anomaly, karyotype and hydrops

	Termination		IUD ¹		Delivery	
	n	%	n	%	n	%
Congenital anomaly						
No	10	29.4	16	47.1	8	23.5
Yes	10	38.5	7	26.9	9	34.6
Karyotype						
Abnormal	9	36.0	10	40.0	6	24.0
Normal	8	38.1	6	28.6	7	33.3
Unknown	3	21.4	7	50.0	4	28.6
Hydrops						
Yes	5	35.7	8	57.1	1	7.1
No	15	32.6	15	32.6	16	34.8

¹Intrauterin death.

21.^[4,15] In first-trimester diagnoses, monosomy X and trisomy 21 had similar prevalence (~20%), whereas in the second trimester, trisomy 21 was more frequent. However, due to the small number of cases diagnosed in the second trimester, statistical comparison was limited. Chromosomal microarray analysis (CMA) and whole-exome sequencing (WES) can detect submicroscopic chromosomal abnormalities and have shown increased diagnostic yield in fetuses with cystic hygroma.^[16] Schreurs et al.^[13] reported that Noonan syndrome was detected in 40% of tested fetuses. Nevertheless, in our center, many patients declined CMA and WES testing primarily due to the financial burden, as these advanced tests are not currently reimbursed by the national health insurance system. This economic limitation restricts equitable access to comprehensive genetic evaluation. Integrating CMA and WES into public healthcare reimbursement policies—particularly for high-risk fetal conditions—may increase diagnostic yield, improve parental counseling, and contribute to more informed decision-making. Congenital anomalies affected 43.3%. Contrary to previous reports that cite cardiac defects as the most common anomaly,^[15,17] abdominal wall defects were most prevalent in our cohort. The presence of congenital anomalies did not significantly differ based on karyotype or gestational age at diagnosis.

The presence of hydrops fetalis is a well-established poor prognostic factor, with previous studies reporting a high mortality rate.^[17–19] In our study, the presence of hydrops was associated with an approximately 87% reduction in the odds of live birth, although this finding was marginally nonsignificant (OR=0.126, p=0.063). This trend aligns with the existing literature, suggesting that hydrops significantly compromises fetal viability. The borderline p-value may be due to the limited sample size, highlighting the need for larger studies to confirm this association. Lymphatic obstruction can cause compression of the aorta and left atrium by causing dysfunction, generalized edema, and fetal death. Similarly, in our study, only one fetus was delivered (7.1%) and survived. This fetus was diagnosed with trisomy 21. In our study, 17 of the patients were delivered, and only 5 of them (8.3%) were alive and well, lower than previous studies in the literature, which report survival rates ranging from 16–18%.^[8,14,13] The cases that were defined as alive and well required no therapy after delivery, and cystic hygroma regressed completely. The other patients who were delivered had structural anomalies or abnormal karyotypes and refused termination. Twelve cases of cystic hygromas (20%) had no structural anomalies and had a normal karyotype. 25% of these cases were terminated (n=3), 33.3% were intrauterine demised (n=4), and 41.7% were delivered and were healthy. Our findings regarding poor perinatal outcomes and high termination rates in fetuses with cystic hygroma are consistent with previous reports on non-immune hydrops fetalis, in which cystic hygromas represent one of the most common identifiable causes and overall survival rates remain low.^[20] Although the prognosis of cystic hygroma is generally poor, it is significantly improved in the absence of unfavorable factors. These results highlight the importance of careful prenatal assessment and counseling in cases of cystic hygroma, as outcomes vary considerably depending on associated anomalies and karyotypic abnormalities. This study has some limitations. The absence of CMA and WES

restricts our ability to detect submicroscopic genetic abnormalities. If these tests had been performed, more genetic pathologies could have been encountered. Additionally, the retrospective, single-center design and modest sample size may limit the generalizability of our findings.

CONCLUSION

Cystic hygroma is strongly associated with chromosomal and structural abnormalities. Pregnancies affected by this condition should receive comprehensive counseling regarding prognosis and be offered karyotyping. Continued monitoring in ongoing pregnancies should include detailed second-trimester ultrasonography and fetal echocardiography. While recurrence data are limited, a thorough prenatal evaluation is recommended in subsequent pregnancies with a history of cystic hygroma.

Disclosures

Ethics Committee Approval: The study was approved by Sakarya University Training and Research Hospital Ethics Committee (No: 2020/500, Date: 07.09.2020).

Informed Consent: Due to the retrospective nature of the study, informed consent has been waived.

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REFERENCES

- Schild RL, Orhan Y, Meyberg H, Braunschweig T, Knöpfle G, Gembruch U. Three-dimensional ultrasound of a massive fetal lymphangioma involving the lower extremity. *Ultrasound Obstet Gynecol* 2003;22:547–9.
- Giacalone PL, Boulot P, Deschamps F, Hedon B, Laffargue F, Viala JL. Prenatal diagnosis of cavernous lymphangioma of the arm. *Ultrasound Obstet Gynecol* 1993;3:48–50.
- Chen CP, Liu FF, Jan SW, Lee CC, Town DD, Lan CC. Cytogenetic evaluation of cystic hygroma associated with hydrops fetalis, oligohydramnios or intrauterine fetal death: the roles of amniocentesis, postmortem chorionic villus sampling and cystic hygroma paracentesis. *Acta Obstet Gynecol Scand* 1996;75:454–8.
- Mack LM, Lee W, Mastrobattista JM, Belfort MA, Van den Veyver IB, Shamshirsaz AA, et al. Are first trimester nuchal septations independent risk factors for chromosomal anomalies? *J Ultrasound Med* 2017;36:155–61.
- Bronshtein M, Zimmer EZ, Blazer S. A characteristic cluster of fetal sonographic markers that are predictive of fetal Turner syndrome in early pregnancy. *Am J Obstet Gynecol* 2003;188:1016–20.

6. Rosati P, Guariglia L. Prognostic value of ultrasound findings of fetal cystic hygroma detected in early pregnancy by transvaginal sonography. *Ultrasound Obstet Gynecol* 2000;16:245–50.
7. Graesslin O, Derniaux E, Alanio E, Gaillard D, Vitry F, Quéreux C, et al. Characteristics and outcome of fetal cystic hygroma diagnosed in the first trimester. *Acta Obstet Gynecol Scand* 2007;86:1442–6.
8. Scholl J, Durfee SM, Russell MA, Heard AJ, Iyer C, Alammari R, et al. First-trimester cystic hygroma: relationship of nuchal translucency thickness and outcomes. *Obstet Gynecol* 2012;120:551–9.
9. Johnson MP, Johnson A, Holzgreve W, Isada NB, Wapner RJ, Treadwell MC, et al. First-trimester simple hygroma: cause and outcome. *Am J Obstet Gynecol* 1993;168:156–61.
10. Noia G, Pellegrino M, Masini L, Visconti D, Manzoni C, Chiaradia G, et al. Fetal cystic hygroma: the importance of natural history. *Eur J Obstet Gynecol Reprod Biol* 2013;170:407–13.
11. Orgul G, Ozyuncu O, Oktem A, Beksac MS. Management and outcomes of cystic hygromas: experience of a tertiary center. *J Ultrasound* 2017;20:127–31.
12. Lajeunesse C, Stadler A, Trombert B, Varlet MN, Patural H, Prieur F, et al. First-trimester cystic hygroma: prenatal diagnosis and fetal outcome. *J Gynecol Obstet Biol Reprod (Paris)* 2014;43:455–62. [Article in French]
13. Schreurs L, Lannoo L, De Catte L, Van Schoubroeck D, Devriendt K, Richter J. First trimester cystic hygroma colli: Retrospective analysis in a tertiary center. *Eur J Obstet Gynecol Reprod Biol* 2018;231:60–4.
14. Kharrat R, Yamamoto M, Roume J, Couderc S, Vialard F, Hillion Y, et al. Karyotype and outcome of fetuses diagnosed with cystic hygroma in the first trimester in relation to nuchal translucency thickness. *Prenat Diagn* 2006;26:369–72.
15. Malone FD, Ball RH, Nyberg DA, Comstock CH, Saade GR, Berkowitz RL, et al. First-trimester septated acystic hygroma: prevalence, natural history, and pediatric outcome. *Obstet Gynecol* 2005;106:288–94.
16. Zhou Y, Lu X, Zhang Y, Ge Y, Xu Y, Wu L, et al. prenatal genetic diagnosis of fetal cystic hygroma: a retrospective single-center study from China. *Cytogenet Genome Res* 2022;162:354–64.
17. Yakıştıran B, Altınboğa O, Canpolat E, Çakar EŞ, Çelen Ş, Çağlar AT, et al. Analysis of cystic hygroma diagnosed in the first trimester: Single-center experience. *J Turk Ger Gynecol Assoc* 2020;21:107–10.
18. Bernard P, Chabaud JJ, Le Guern H, Le Bris MJ, Boog G. Cystic hygroma of the neck. Antenatal diagnosis, prognostic factors, management. 42 cases. *J Gynecol Obstet Biol Reprod (Paris)* 1991;2:487–95.
19. Özcan HÇ, Uğur MG, Balat Ö, Sucu S, Bayramoğlu Tepe N, et al. Analysis of cystic hygroma diagnosed in the prenatal period: 5-yearsı experience at a tertiary hospital in Southeastern Turkey. *J Matern Fetal Neonatal Med* 2019;32:1800–5.
20. Erdoğan E, Arısoy R, Uygur L, Sancak S, Kayabasoğlu F, Arıkan SA, et al. Perinatal outcomes of 91 cases of non-immune hydrops fetalis. *Zeynep Kamil Med J* 2022;53:163–9.