Ref: Ro Med J. 2021;68(Suppl5)
DOI: 10.37897/RMJ.2021.S5.16

Spontaneous regression of large fetal antero-lateral neck cysts: A case report and literature review

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ABSTRACT

The entity of fetal lateral cervical cysts is presented as an isolated anomaly or associat-ed with other sonographic structural findings, sometimes being determined by chromo-somal abnormalities. We report a case of spontaneous regression of large fetal anterol-ateral cervical bilateral cysts diagnosed by ultrasonography at 13 weeks. Complete res-olution of cervical cysts was documented at 20 weeks. The patient reveals no concern-ing results to other assessments related to her pregnancy including first trimester bio-chemical screening, cell free fetal DNA testing, routine laboratory tests, ultrasound re-peated evaluation of fetal growth and wellbeing. When fetal anterolateral cervical cysts are encountered as isolated finding, completely separated by nuchal translucency fluid collection, regardless of the cystic diameter and bilaterally, the rules are normal karyo-type, high rate of total regression and preservation the good pregnancy outcome.

Keywords: anterolateral cervical cyst, fetal lateral neck cyst, jugular lymphatic communication se-quence

INTRODUCTION

The evolving techniques of performing ultrasonography scans as early assessment of the fetus, reveal structures that are difficult to be diagnosed as physiological or pathological variant and may be challenging to manage. The finding of lateral cervical cysts poses questions regarding additional invasive or non-invasive investigation, follow-up and prognostic. As far as we know, fetal lateral cervical cysts are rare, but studies from the literature report an incidence ranging from 2.4% to 6.9%, which raises the issue of the existence of a genetic predisposition and specific risk factors responsible for a particular geographical distribution of this pathologic entity [1-5]. Among these cases, isolated cysts have better outcome, compared to non-isolated cysts where association with nuchal translucency over 3 mm increases the risk for fetal aneuploidy [1-3,5].

CASE PRESENTATION

Pregnant patient of 34 years old presents for fetal first trimester screening at 13 weeks of gestation. She is gravida II, para II, has no abortions but one previous uncomplicated pregnancy and no relevant medical, family or surgical history; her husband does not has any medical or genetic particularity. At the first visit she weighed 83 kg, her height was 162 cm, her blood group was OI, RhD positive, being in

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Article History:
Received: 4 November 2021
Accepted: 29 November 2021

incompatibility state without alloimmunization with her husband's blood group B and RhD positive. The patient was on no medication other than pregnancy supplements. During first trimester ultrasound screening performed by a specialist in maternal fetal medicine, two large fetal antero-lateral cervical avascular cysts, measuring in longitudinal diameter 7.3 mm and 8.3 mm, respectively, without communication to nuchal translucency have been found (Figures 1-3). Nuchal translucency was in appropriate range for gestational age, 2.50 mm (Figure 4), and no other structural anomaly or soft marker for chromosomal anomalies has been notice. The cell-free fetal DNA (cffDNA) test sustain that no aneuploidies of chromosomes 21, 13, 18, 16, 9 or sex chromosomes were identified. The results show the presence of a diploid number for chromosomes 21, 18, 13, 16, 9, with two sex chromosomes (XY); no investigated microdeletions related syndromes (22q11.2 deletion in Velo cardio facial syndrome, 1p36 deletion syndrome, 15q11.2 deletion in Angelman syndrome, 15q11.2 deletion in Prader-Willi syndrome, 5p- deletion in Cri du Chat syndrome, or 4p- deletion in Wolf-Hirschhorn syndrome) were detected.

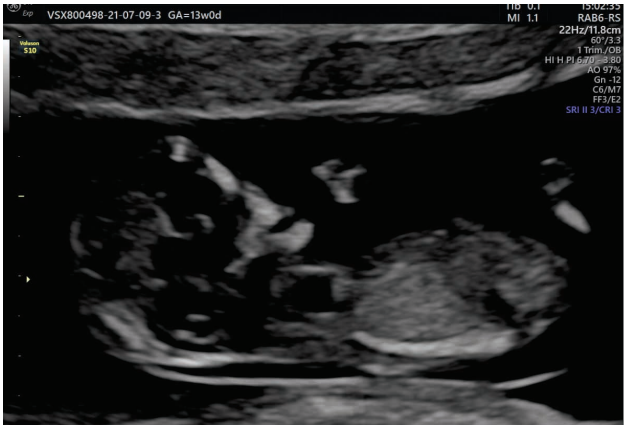

FIGURE 1. Ultrasound image of fetal parasagittal section at 13 weeks of gestation used to evidence the location of the bilateral cervical cysts and spatial relation with the nuchal translucency

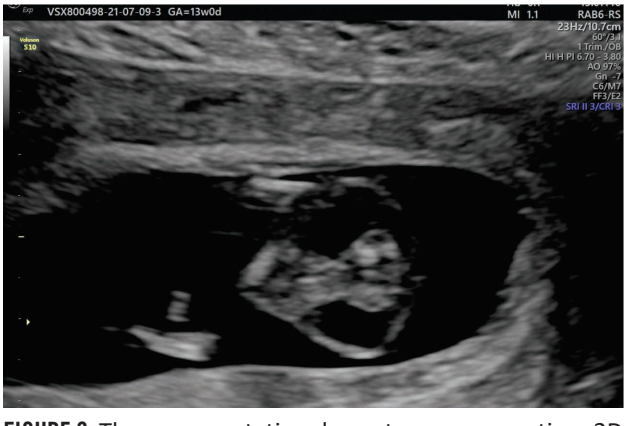

FIGURE 2. The same gestational age, transverse section, 2D imaging: anterolateral cysts of the fetal neck do not communicate posteriorly

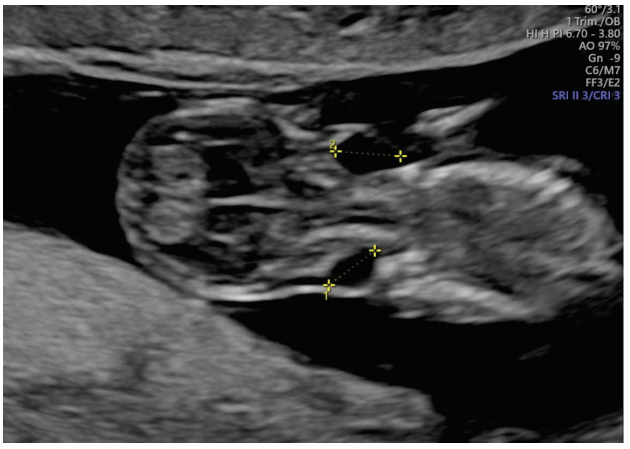

FIGURE 3. Frontal section showing large antero-lateral bilateral cervical cysts measuring 8.30 mm and respectively 7.30 mm in the longest diameter

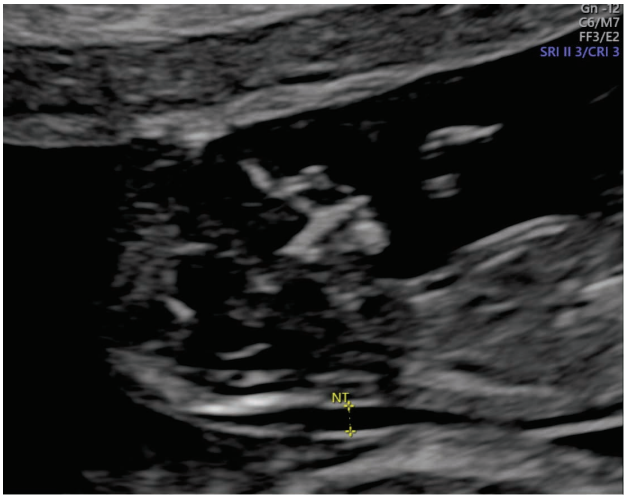

FIGURE 4. Ultrasound measuring of fetal nuchal translucency on sagittal standard section: 2.50 mm at 13 weeks of gestation

Three weeks later, decreased diameters of anterolateral cervical cyst measuring 5.1 mm and respectively 4.6 mm (Figure 5) and no other new developed fetal structural anomalies have been found by specialized ultrasound. The reduction of the cists diameters, along with normal laboratory and ultrasonography results, were reassuring for the transient evolution of these cysts that seemed to be more functional than structural anomaly that temporarily disturbed developmental process.

At 20 weeks scan complete resolution of antero-lateral cervical cysts was observed. The spontaneous remission was maintained within the seriated scans that assessed the pregnancy evolution until birth. The patient was advised to continue routine medical visits until delivery, and she received the information about the good outcome that is expected with spontaneous remission of isolated fetal neck cysts, in the context of physiological course of her pregnancy: appropriate biometry for gestation-

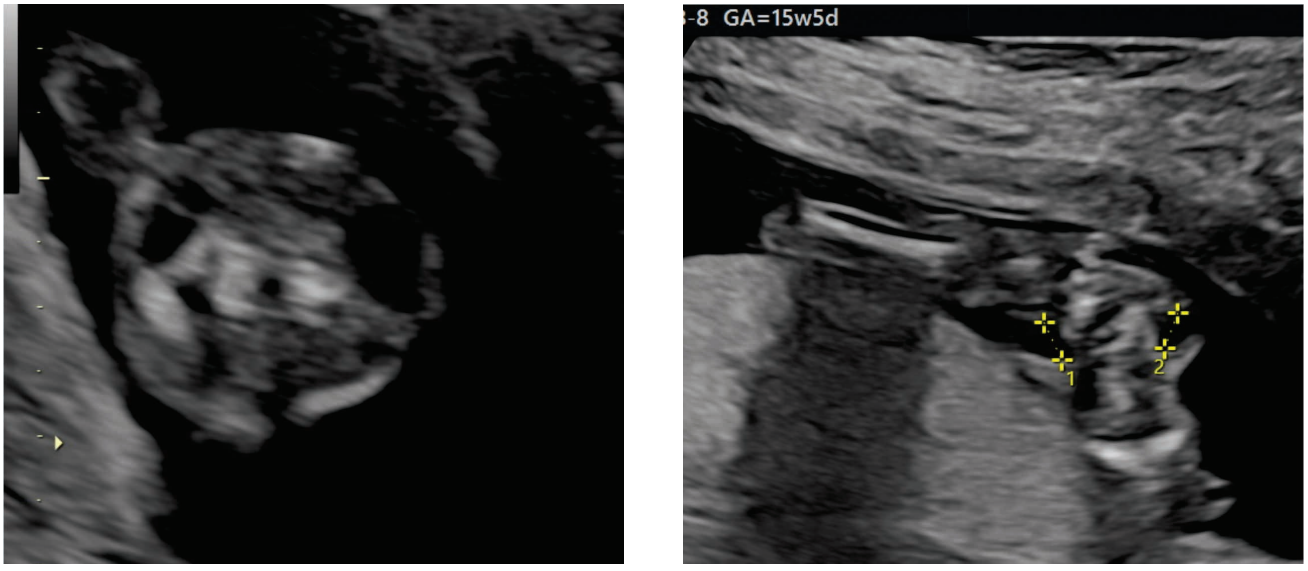

FIGURE 5. Ultrasonographic image of fetal neck in transverse section at 15 weeks and 5 days, showing a decrease in anterolateral cervical cysts diameter to 5.10 mm and respectively 4.60 mm

al age, normal Doppler studies, a good score on fetal biophysical profile. If the fetal anterolateral cysts would persist, we would recommend imagistic evaluation by fetal MRI. An abnormal fetal DNA results would require invasive tests, respectively amniocentesis for karyotyping optimal by microarray.

The patient was given the informed consent for anonymous publishing her case.

DISCUSSION

Fetal neck cysts may be delineated into three main categories: the spectrum of cystic hygroma colli, embryonic remnants as a persistent structure and functional cysts with spontaneous regression. In our case, we were confronted with differential diagnosis for: cystic hygroma colli, which is situated at the level of fetal neck but orientated posteriorly, jugular lymphatic communication sequence representing a physiological delay that can induces un increased risk for fetal aneuploidy if is associated with larger nuchal thickness, partial or complete pathological obstruction of drainage pathways, nuchal edema, fetal hydrops, branchial, thymic and parathyroid anomalies [6,7]. A careful searching for other fetal structural anomalies should be performed because lateral neck cysts points also to a greater probability of heart malformation [3].

Many authors agree that the finding of cervical cysts should be followed by fetal karyotyping even in those cases of isolated cysts because abnormal results has been reported even in apparent normal fetuses [1,2,4], while others consider that no additional work up is needed in case of isolated cysts [3]. In the context of the absence of any other structural and soft markers anomalies, we have considered reassuring the extensive cell-free fetal DNA test.

Some data available in literature consider that cysts diameter around or smaller than 3 mm indicates a better outcome even in the presence of increase nuchal translucency, but other sources show no relevance of cyst size or bilateral findings in the terms of outcome [2,3].

CONCLUSIONS

Our case is most probably representative for physiological delay of jugular lymphatic communication despite large diameter of the cysts, thus we support the conclusion of some authors that cyst diameter is not link to adverse outcome, but we strongly advise for non-invazive prenatal test as alternative for first trimester biochemical screening and as first step for genetical investigation of all minor or major anomalies.

Based on favorable prognostic indicators like normal nuchal translucency thickness, no other malformations depicted and normal karyotype, most probably this finding is due to physiological delay of jugular lymphatic communication system, and we confirm the excellent outcome of these cases.

Conflict of interest: none declared
Financial support: none declared

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