

Congenital malformations: case reports of routine use of obstetrics ultrasound

FULVIA BUOGO & IGNAZIO SPARACIO

via V. Di Marco 43, 90143 Palermo, Italy

SUMMARY

Ultrasounds in medicine are a simple and effective instrumental system of investigation in the diagnosis and monitoring of numerous pathologies. In particular, the routine use of this method is fundamental in obstetrics outpatient for the control of pregnancies. Periodic ultrasound controls allow to follow the normal embryo-fetal growth and to identify any defects for early prenatal diagnosis. In this paper we refer 5 case reports of congenital malformations highlighted during outpatient ultrasound scans of women in pregnancy.

KEY WORDS

Congenital malformations; obstetrics ultrasound; prevention.

Received 18.12.2015; accepted 08.02.2016; printed 28.02.2016

INTRODUCTION

Prenatal diagnosis is a complex of instrumental and laboratory investigations aimed at assessing and monitoring the embryo-fetal health during all stages of pregnancy.

It allows early detection of chromosomal, infectious hematological, morphological and pathophysiological diseases and the identification of possible risk factors for the mother and fetus by promoting

better medical management of pregnancy and childbirth in relation to the highlighted clinical conditions.

The methods of prenatal diagnosis are divided into invasive (chorionic villus sampling, amniocentesis, cordocentesis, ...) and non-invasive (fetal ultrasound, biochemical and molecular studies on the mother's blood, ...).

Non-invasive methods include, above all, the ultrasound examination which is particularly important in gynecology and obstetrics surgery, either external or within hospital.

The ultrasound scans during pregnancy allow to highlight the normal fetal growth and to point out possible birth defects. There is a large international series of studies about this topic and specific guidelines are regularly updated (GRANDJEAN ET AL., 1999; STOLL ET AL., 1995; ISUOG, 2013; SIEOG, 2015).

A malformation is a morphological defect of an organ or a part of it, or of more or less extensive areas of the body resulting from an intrinsically abnormal process of development in the early stages of growth. Congenital malformations are divided into major, when they have a clear medical and/or surgical importance, with obvious consequences on morbidity and mortality of the interested individual, and minor, when they have less clinical significance and do not appreciably affect the duration and quality of life.

The ultrasound diagnosis of these congenital anomalies is very important and used; it is based essentially on the recognition of a pathological deviation of the normal-fetal anatomy. The first studies on this topic have been published by DONALD & BROWN (1961), SUNDEN (1964) and CAMPBELL ET AL. (1972) with a number of other contributions that have followed over the years until today (see also DEUTCHMAN ET AL., 1997; SPEARS & GRIFFIN, 1999; CAREY, 2000; GOLDSTEIN, 2002; BORRELLI ET AL., 2008; CORSELLO & GIUFFRÈ, 2012).

In this paper we refer 5 case reports of congenital malformations highlighted during outpatient ultrasound scans of women in pregnancy.

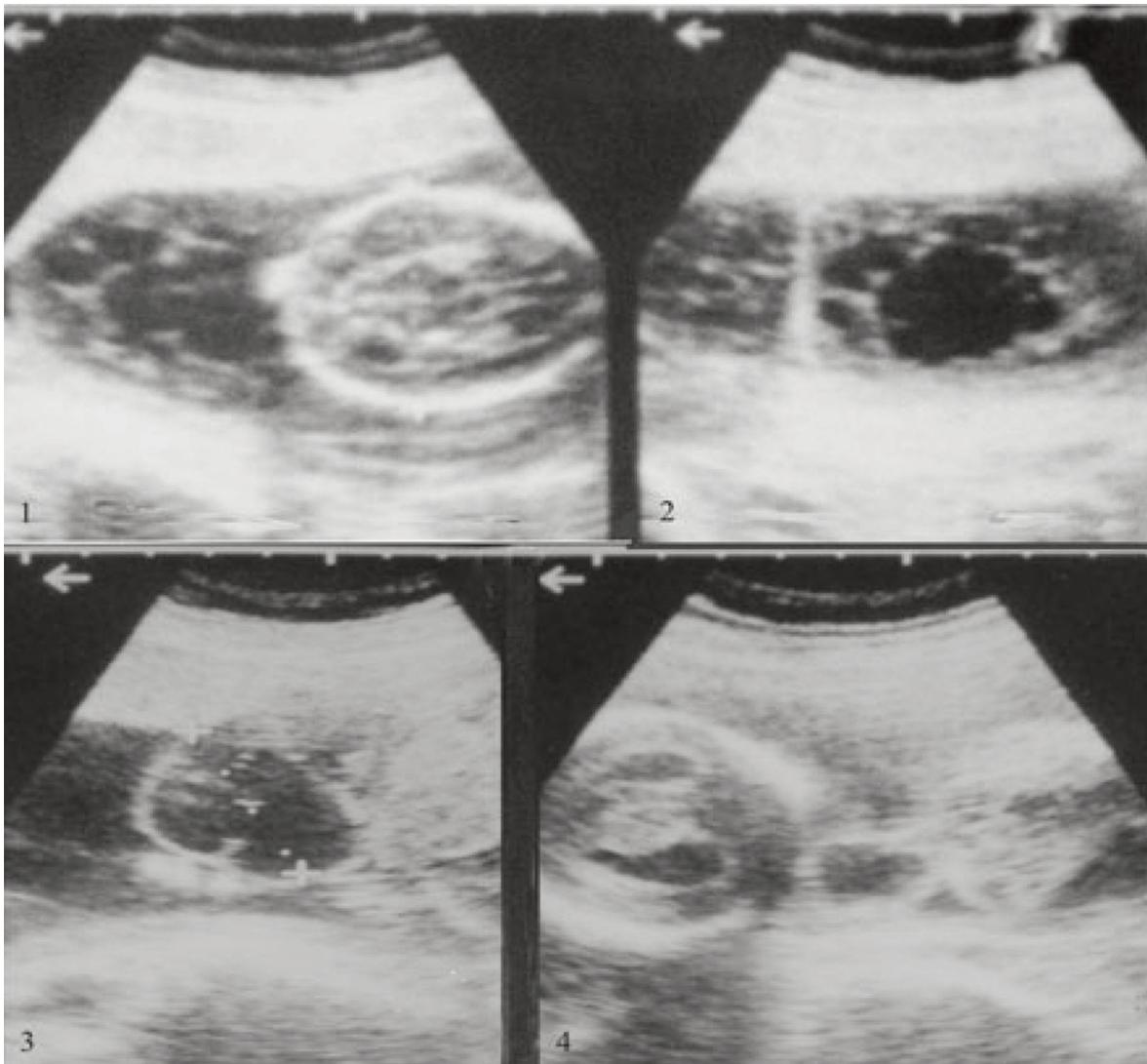
CASE REPORTS

Case 1

Woman 32 years at 1st pregnancy. Clinical monitoring and ultrasound check Q1 not exhibited. She comes to our observation at 20 weeks of amenorrhea for clinical monitoring and ultrasound. It is performed a diagnosis of cystic deformation of the neck compatible with a diagnosis of cystic hygroma; with

also bilateral hydrocephalus and ascites. After hospitalization and confirmation of the diagnosis, therapeutic abortion was performed.

The cystic hygroma of the neck is caused by non-union of lymphatic vessels, usually, laterocervical. This obstruction leads to dilation of the local structures with formations of cystic areas. Usually this pathology is localized in the back of the neck but also on the thorax and abdomen. The cystic hygroma can usually be isolated or be part of more complex syndromes.



Figures 1–4. Ultrasound diagnosis of fetal malformations (see text). Figs. 1–3. Cystic hygroma of the neck. Figure 4. In the same fetus is evident the presence of bilateral hydrocephalus.

Case 2

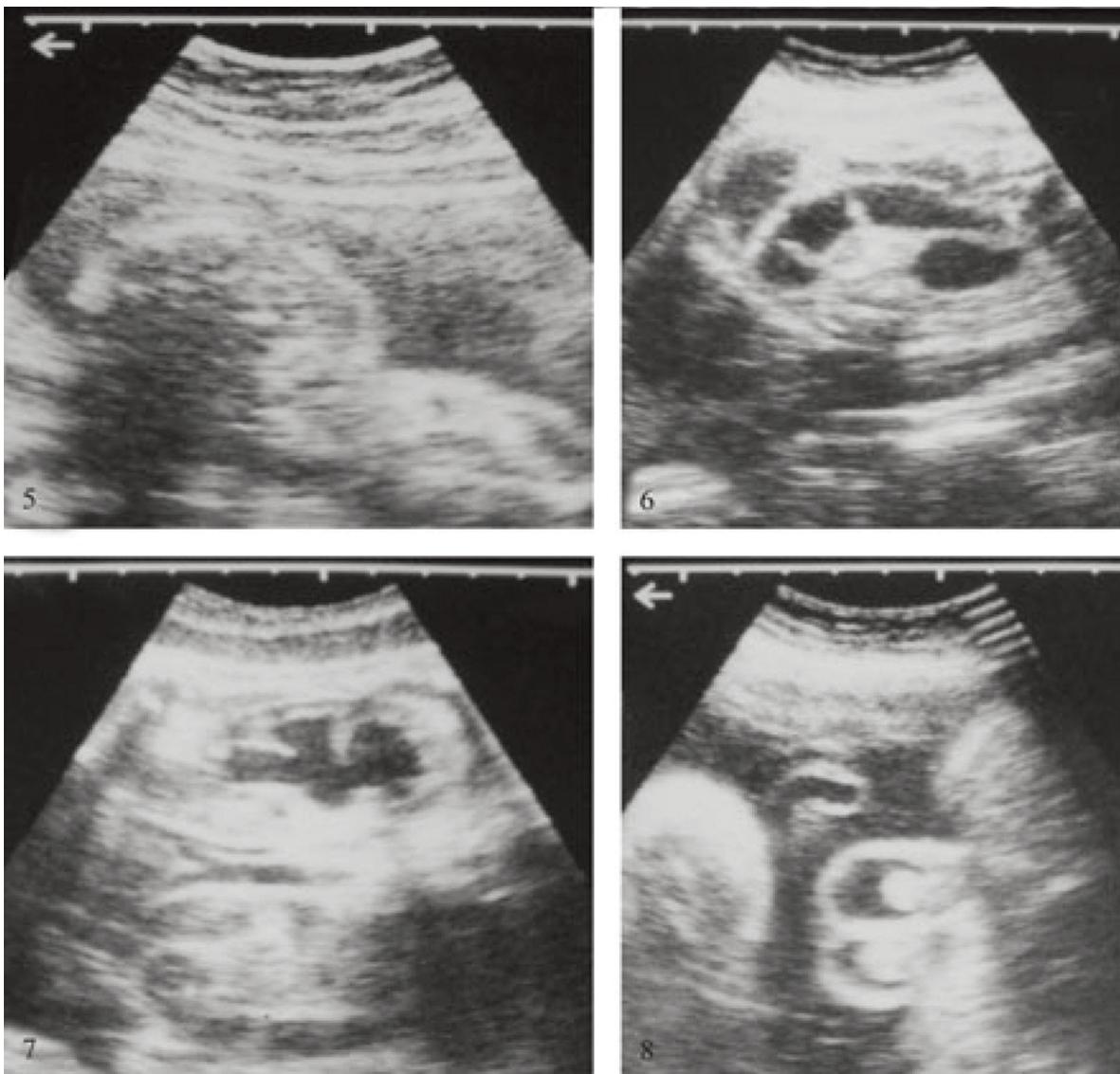
Woman 33 years at 1st pregnancy. Control outpatient ultrasound at 22 weeks. She had not carried out the Q1 checks. Diagnosis of anencephaly was confirmed at the next hospitalization when she performed therapeutic abortion.

The anencephaly consists in the absence, more or less complete, of the cranial vault with the brain tissue that, although present, is degenerated due to the lack of adequate bone protection. Ultrasound

diagnosis is easy because the fetal head can be identified in its normal anatomy from the very first weeks of pregnancy.

Case 3

Woman 34 years at 2nd pregnancy. Normal pregnancy until ambulatory monitoring at 34 weeks of gestation, when at the ultrasound there is a growth retardation of three weeks on the scheduled date; the



Figures 5–8. Ultrasound diagnosis of fetal malformations (see text). Fig. 5. Anencephaly. Fig. 6. Intestinal obstruction. Fig. 7. Renal pelvis dilatation. Fig. 8. Bilateral hydrocele.

intestinal loops are stretched with the presence of significant amount of fluid and corpusculated material in suspension. Diagnosis of suspected intestinal obstruction. The ultrasound performed after three weeks confirmed the diagnosis. Birth at the end of the 39 weeks with the baby in good general conditions, mild respiratory distress, weight 2,400 kg, meteoric abdomen with normal anal orifice. The day after the birth he is operated in urgency for bowel obstruction (volvulus) with initial perforation. A regular post-operative course followed and completed "restitutio ad integrum".

Case 4

Woman 29 years at 2nd pregnancy. Normal pregnancy until the visit at 39th week of amenorrhea when the ultrasound was performed with a diagnosis of renal pelvis dilatation at the right kidney. Birth at 39th week with child in good conditions; diagnosis of right kidney hydronephrosis - which normalized after one month from birth - was confirmed.

Case 5

Woman 30 years at 2nd pregnancy. Normal pregnancy until ambulatory monitoring after 36 weeks of amenorrhea, when the ultrasound is performed with a diagnosis of bilateral hydrocele. The photography section highlights the clear division in the two emisroti where the testicles have already descended. These appear to be more highlighted because of fluid collection (hydrocele) around them.

Childbirth normal. Confirmed hydrocele at birth and called "paraphysiological".

REFERENCES

- BORRELLI A.L., ARDUINI D., CARDONE A. & VENTRUTO V., 2008. Medicina dell'età prenatale. Prevenzione, diagnosi e terapia dei difetti congeniti e delle principali patologie gravidiche. Springer-Verlag Italia, Milano, 581 pp.
- CAMPBELL S., HOLT E.M., JOHNSTONE F.D. & MAY P., 1972. Anencephaly: Early ultrasonic diagnosis and active management. *Lancet*, 2: 1226–1227.
- CAREY J.C., 2000. Integrating prevention into obstetrics/gynecology. *Academic Medicine*, 75 (7 Suppl.): S72–76.
- CORSELLO G. & GIUFFRÈ M., 2012. Congenital malformations. *Journal matern Fetal Neonatal Medicine*, 25 (Suppl. 1): 25–29. Doi: 10.3109/14767058.2012.664943. Epub 2012 mae 14
- DEUTCHMAN M.E. & HAHN R., 1997. Office procedures Obstetric ultrasonography. *Primary Care*, 24: 407–431.
- DONALD I. & BROWN T.G., 1961. Localization using physical devices radioisotopes and radiographic methods, I: Demonstration of tissue interfaces within the body by ultrasonic echo sounding. *British Journal of Radiology*, 34: 539–546.
- GOLDSTEIN S.R., 2002. Routine use of office gynaecologic ultrasound. *Journal of Ultrasound in Medicine*, 21: 489.
- ISUOG, 2013. Practice Guidelines (updated): sonographic screening examination of the fetal heart. *Ultrasound in Obstetrics and Gynecology*, 41: 348–359.
- GRANDJEAN H., LARROQUE D. & LEVI S., 1999. The performance of routine ultrasonographic screening of pregnancies in the Eurofetus Study. *American Journal of Obstetrics and Gynecology*, 181: 446–454.
- SIEOG, 2015. Linee Guida. Società Italiana di Ecografia Ostetrico Ginecologica Edizione 2015. Editeam, Gruppo Editoriale, Roma, 168 pp.
- SPEARS G. & GRIFFIN J., 1999. Ultrasound in obstetric triage. *Journal of Nurse-Midwifery*, 480.
- STOLL C., DOTT B., ALEMBIK Y. & ROTH M.P., 1995. Evaluation of routine prenatal diagnosis by a registry of congenital anomalies. *Prenatal Diagnosis*, 15: 791–800.
- SUNDEN B., 1964. In the diagnostic value of ultrasound in obstetrics and gynecology. *Acta Obstetric Gynecology Scandinavia*, 43: 121–123.