Ventriculomegaly

7

Ventriculomegaly means enlargement of the lateral ventricles. Ventriculomegaly is diagnosed when the width (transverse dimension) of the atrium of the lateral ventricle is 10 mm or more in the second and third trimesters. It is classified as mild (10-12 mm), moderate (13-15 mm) and severe (> 15 mm).

7.1 Mild & Moderate Lateral Ventriculomegaly

Mild or moderate lateral ventriculomegaly is a finding or a sign of a possible disorder and not a disorder by itself. Hence ventriculomegaly is not the final diagnosis. It prompts further investigation to make the final diagnosis. Visualisation and measurement of the near lateral ventricle will determine whether the ventriculomegaly is unilateral or bilateral, symmetric or asymmetric. Once ventriculomegaly is identified, one must perform a dedicated fetal neurosonogram. The highest possible frequency transducer should be used. Depending on the position of the fetal head, one may use transabdominal or transvaginal routes. Acquiring and storing 3D volumes enable application of image enhancing algorithms and display in multiplanar and render modes. In select cases, MRI is indicated to confirm ultrasound findings and to detect additional abnormalities, if any. MRI is useful in cases where ultrasound examination is suboptimal due to maternal habitus, fetal position or advanced gestational age. A thorough global fetal examination to detect any extracranial associated findings is necessary. Mild and moderate lateral ventriculomegaly are termed 'isolated' if there are no other abnormal intracranial or extracranial findings after a thorough examination.

The following practical checklist helps to detect additional findings:

1. CSP: Presence or absence, shape and size
   CSP abnormality is the lead sign for the detection of holoprosencephaly and callosal and septal disorders.
2. Lateral ventricular margin: Smooth and regular or crinkled and irregular
   Irregular lateral ventricular margins are seen in periventricular nodular heterotopia.
3. Choroid plexus: Compare size and echopattern on either side
   Large choroid plexus is seen in intraventricular hemorrhage and choroid plexus papilloma.
4. Lateral ventricular fluid content: Clear or turbid, presence or absence of clot, band or synechium
   Turbid fluid and synechiae are seen in intraventricular hemorrhage or infection.
5. Corpus callosum: Presence or absence and length and thickness
   Failure to visualise a part of or the whole CC indicates partial or complete agenesis of CC, respectively.
6. Cerebral hemispheres: Compare the volume of the two hemispheres
   Asymmetric hemispheric size is seen in unilateral cerebral hypoplasia/atrophy and hemimegalencephaly.
7. Subarachnoid space: Normal, increased or decreased
   The subarachnoid space is obliterated in obstructive hydrocephalus. It is increased in cerebral hypoplasia/atrophy.
8. Cerebral parenchyma:
   (a) Abnormal thinning (cerebral atrophy)
   (b) Presence of cleft(s) (schizencephaly)
   (c) Delayed or premature sulcation (lissencephaly or polymicrogyria, respectively)
   (d) Periventricular hyperechoic halo (infection), calcification or cyst (infection) or solid nodules (periventricular heterotopia)
   (e) Parenchymal solid or cystic lesion (tumor, bleed or cyst).
9. Posterior cranial fossa: Shape, size and position (rotation) of the vermis, size and symmetry of cerebellar hemispheres and presence and size of the cisterna magna
   Mega cisterna magna, Blake's pouch cyst, vermian hypoplasia or Dandy-Walker malformation can be identified.

B. S. Rama Murthy, *Imaging of Fetal Brain and Spine*, https://doi.org/10.1007/978-981-13-5844-9_7

10. Doppler evaluation: Useful in certain cases.
    Abnormal circle of Willis in HPE, relation of suprasellar tumor to the circle of Willis and abnormal course of pericallosal artery in complete agenesis of the corpus callosum are some examples.
11. Targeted anomaly scan to assess:
    (a) Markers of chromosomal abnormality
    As a marker of trisomy 21, the likelihood ratio for isolated mild lateral ventriculomegaly is 3.81.
    (b) Associated findings of single gene syndromes such as Walker-Warburg, Joubert, Meckel, hydrolethalus, Bardet-Biedl (Fig. 7.1a, b).
    (c) Associated findings in fetal infection including hepatomegaly, speckled hepatic hepatic calcification, cataract, IUGR, ascites and hydropic changes.
    (d) Other sporadic anomalies. Fetal echocardiography is indicated.
12. Fetal karyotyping / chromosomal microarray (CMA) and workup for fetal infection. Decision for invasive testing (amniocentesis) would depend on the presence of other abnormalities or soft markers and the first-trimester combined test aneuploidy risk assessment (Figs. 7.2 and 7.3). CMA enables diagnosis of microdeletions or duplications and is best done in consultation with a geneticist.

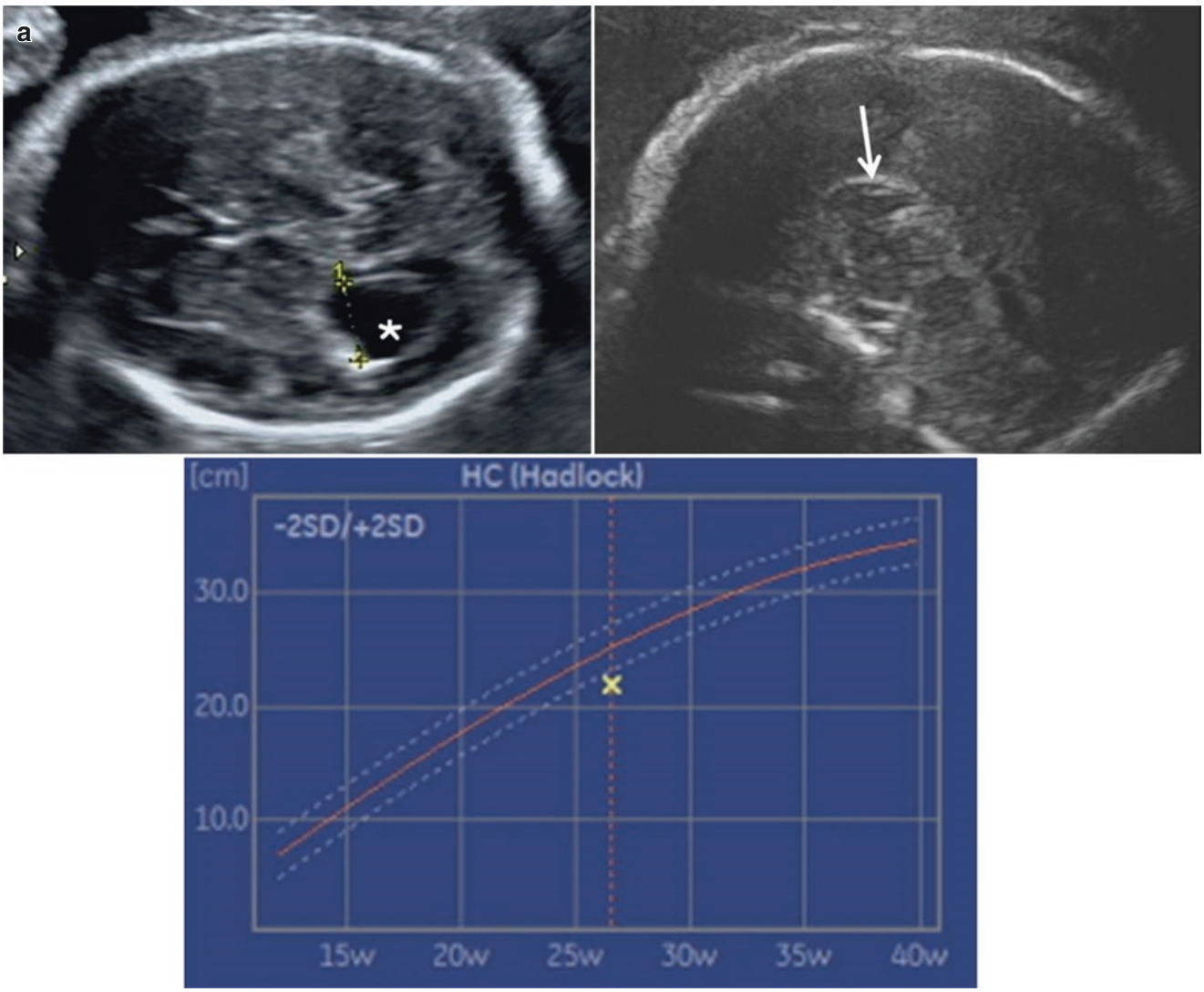


Fig. 7.1 (**a**) 26 weeks (TAS) *microcephaly, partial agenesis of the corpus callosum with dysmorphic facies* in a case of previous child with *Seckel syndrome* – transventricular and midsagittal sections – bilateral mild lateral ventriculomegaly (*), partial callosal agenesis (posterior body and splenium are absent) (solid arrow), HC at −3SD. (**b**) 26 weeks (TAS, 3D US, postnatal MRI of the previous child) *microcephaly, partial agenesis of the corpus callosum with dysmorphic facies* in a case of previous child with *Seckel syndrome* – facial profile and 3D surface rendering of fetal face, picture of the face of previous child, T2W midsagittal cranial section of previous child – sloping forehead (solid arrow), partial callosal agenesis (dotted arrow). Microcephaly, intracranial findings and facial dysmorphism in the fetus are indicative of recurrence of Seckel syndrome (autosomal recessive)

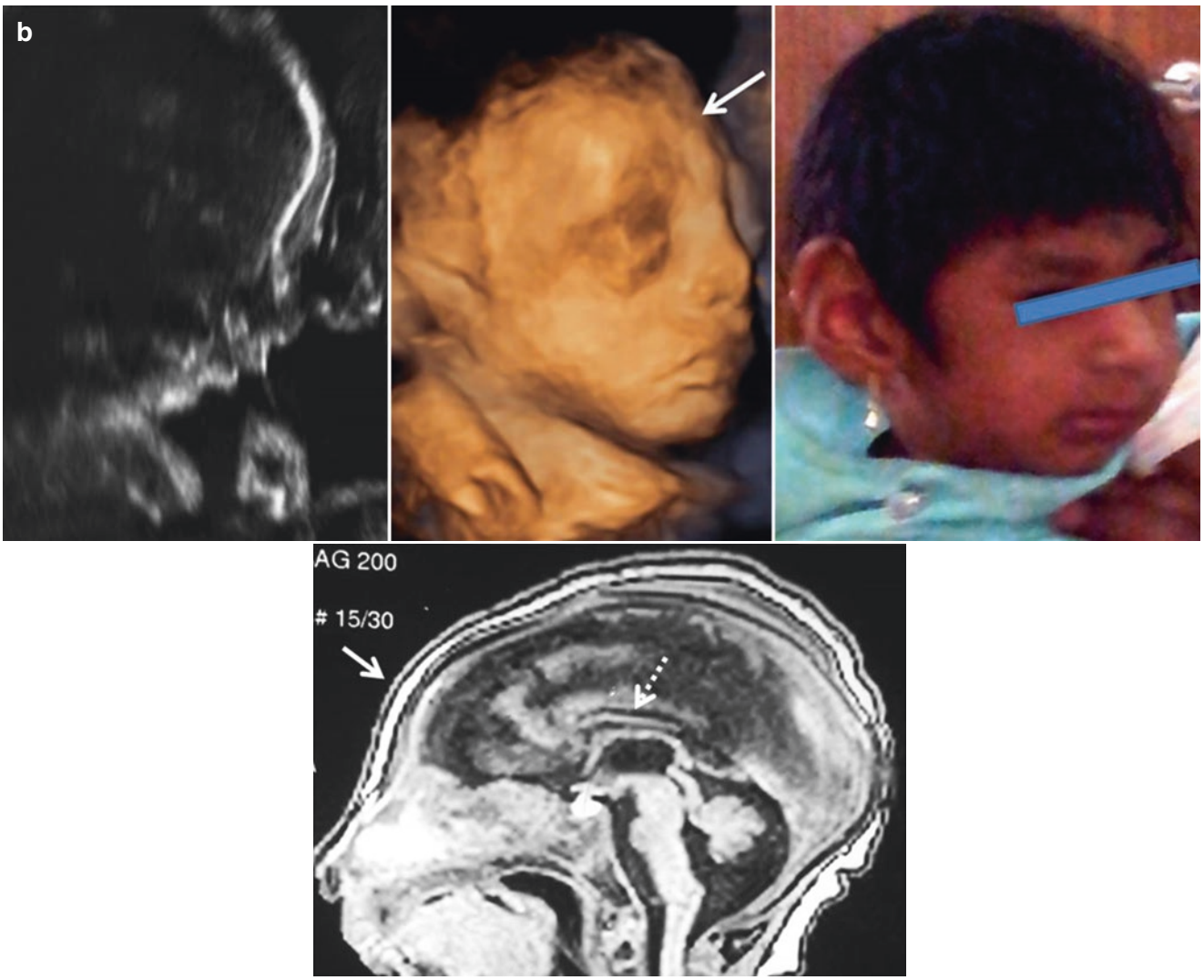


Fig. 7.1 (continued)

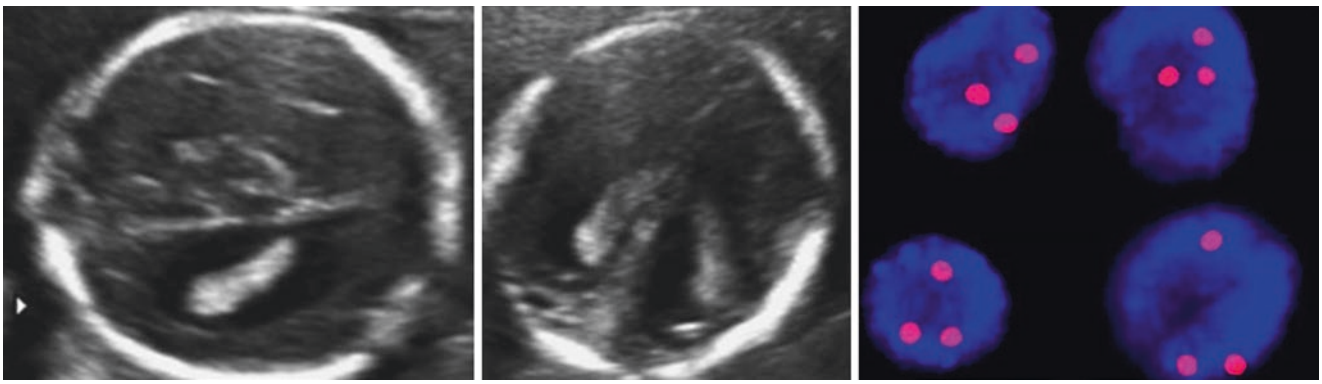


Fig. 7.2 18 weeks (TAS) *Down syndrome* – axial transventricular axial sections and FISH picture – bilateral mild lateral ventriculomegaly with screen-positive triple marker test, amniotic fluid interphase FISH is positive for trisomy 21 (three red signals per amniocyte), karyotype confirmed trisomy 21

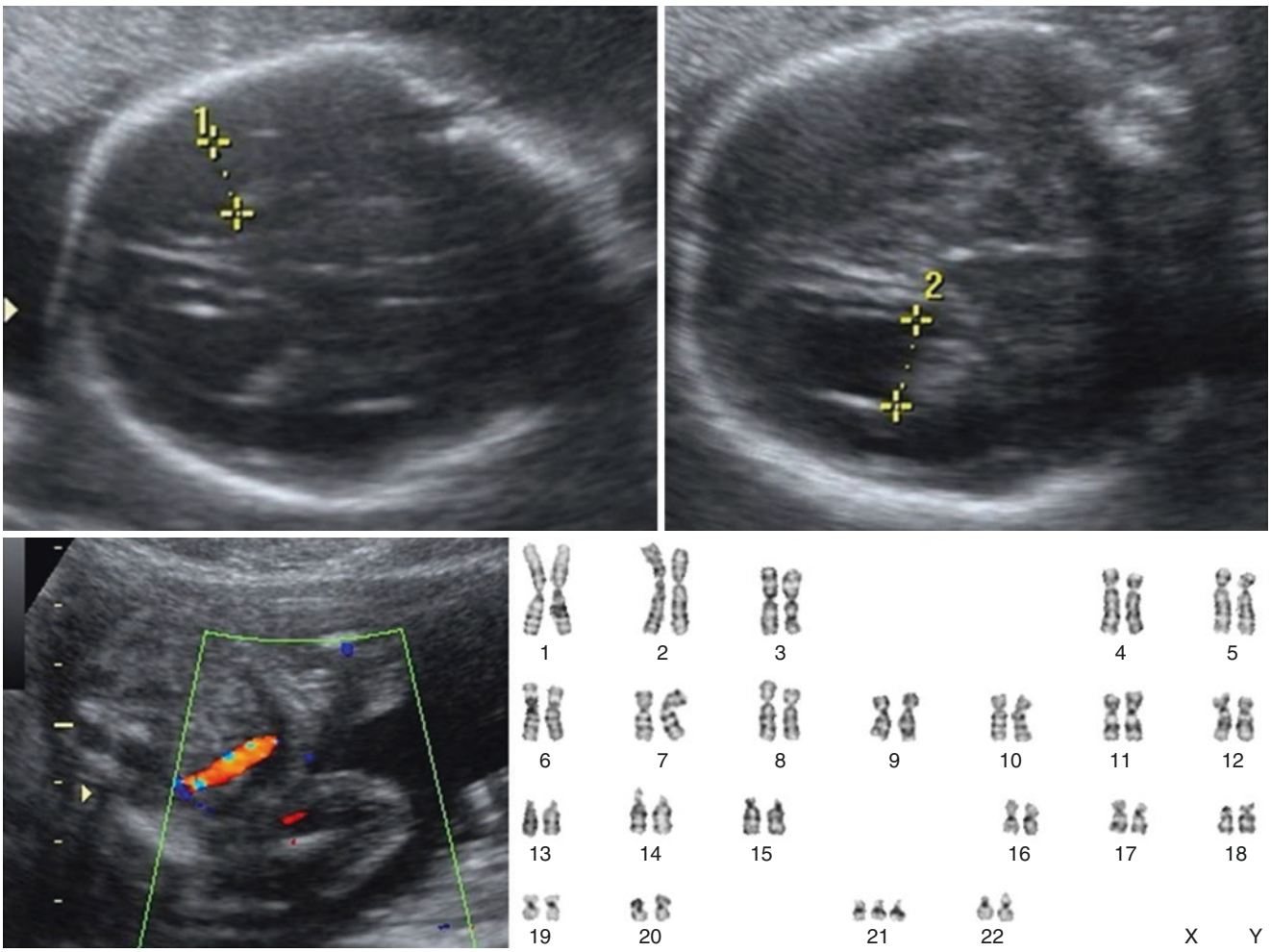


Fig. 7.3 21 weeks (TAS) *Down syndrome* – axial transventricular sections, color Doppler of umbilical artery and karyotype – bilateral mild lateral ventriculomegaly with single umbilical artery, amniotic fluid karyotype is positive for trisomy 21

13. The potential for fetal MRI to detect additional findings depends on the degree of lateral ventriculomegaly as well as the quality of US examination (whether a detailed neurosonogram was performed by an operator with requisite skill and expertise).
14. About 5% of cases with mild and moderate ventriculomegaly are due to infective causes (CMV and toxoplasma). Lateral ventricular dilatation occurs due to gliosis and aqueductal stenosis or cerebral atrophy and ex vacuo phenomenon. Testing amniotic fluid for CMV and Toxoplasma by PCR is an accurate test for fetal infection. However it has a lower sensitivity (45-80%) when tested prior to 21 weeks as compared to a sensitivity of 97-100% when tested after 21 weeks. This is due to the fact that the fetus excretes the virus/protozoal proteins into the amniotic fluid about 6 weeks after the primary infection. In women who decline amniocentesis, serum testing for IgM and IgG antibodies with IgG avidity for CMV and toxoplasma may be done. If negative, infection is ruled out. High avidity indicates remote infection.
15. Repeat ultrasound examination for lateral ventricular size and sulcatory progress after 3–4 weeks interval. Increasing lateral ventricular size is seen in obstructive hydrocephalus. Delayed sulcation is seen in lissencephaly.

Incomplete neurosonography, failure to follow the checklist, inadequate targeted scan for fetal anomalies and occurrence of late-onset anomalies are the reasons for missing associated intra- or extracranial abnormalities. Approximately 7 to 10% of fetuses with apparently isolated mild lateral ventriculomegaly are found to have structural abnormalities after birth. In truly 'isolated' bilateral mild lateral ventriculomegaly, the incidence of abnormal neurodevelopmental outcome is 11%. In truly 'isolated' unilateral mild lateral ventriculomegaly, the incidence of abnormal neurodevelopmental outcome is 7% (Fig. 7.4). Progressive mild lateral ventriculomegaly is associated with abnormal neurodevelopmental outcome in 16% of cases.

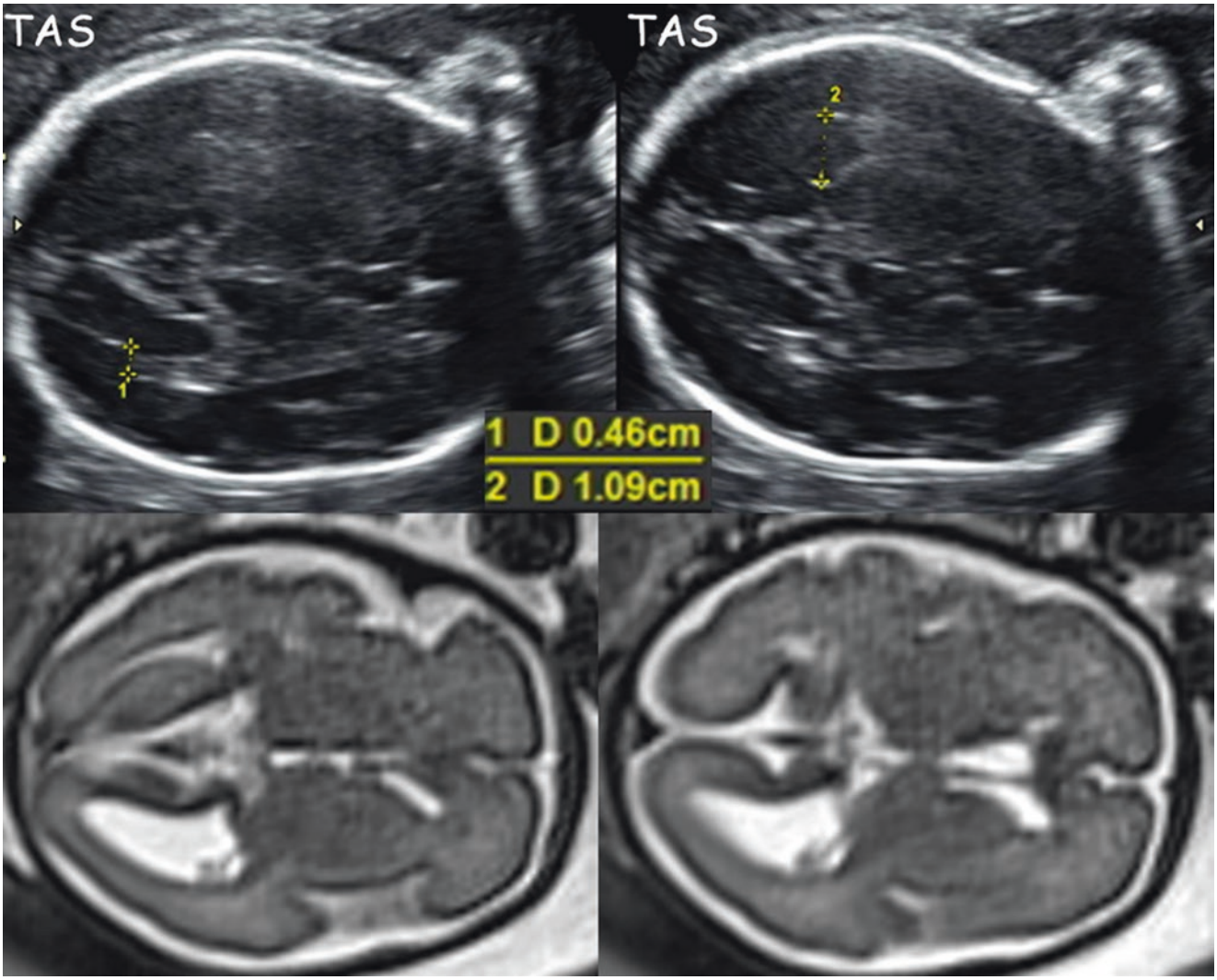


Fig. 7.4 28 weeks (TAS, TVS and MRI) *unilateral lateral ventriculomegaly with normal outcome* – US and T2W axial, coronal and sagittal sections – unilateral mild left lateral ventriculomegaly, sulcation is appropriate for gestational age, the corpus callosum is normal, no periventricular solid/cystic nodules or calcification, posterior cranial fossa is normal. No extracranial anomalies were noted. No increase in the ventriculomegaly noted at 32 weeks. Maternal infection serology and fetal karyotype were normal. This, therefore, is isolated unilateral lateral ventriculomegaly. Postnatal follow-up of 3 years was normal

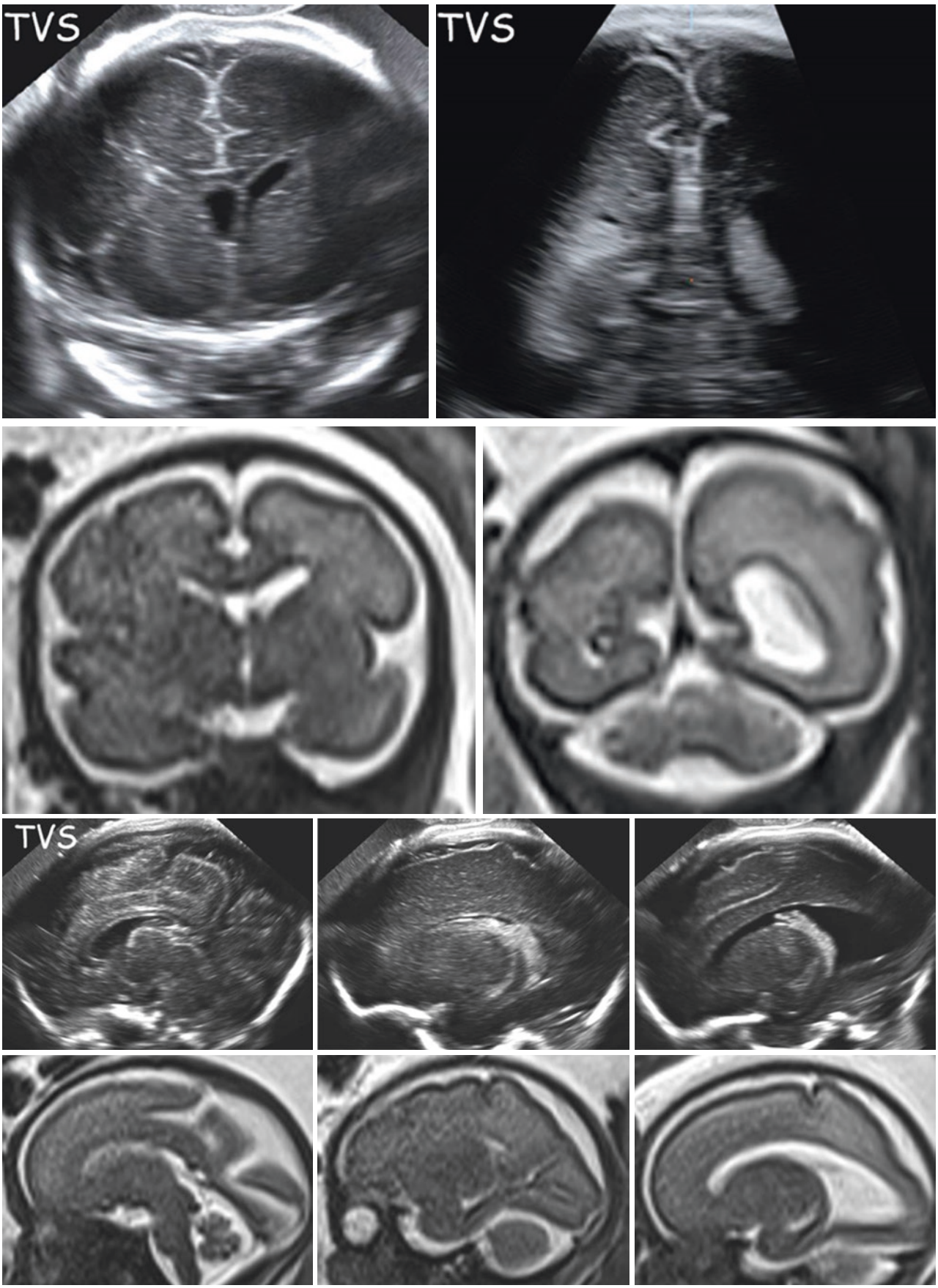


Fig. 7.4 (continued)

7.2 Severe Lateral Ventriculomegaly

Lateral ventricular atrial width equal to or more than 15 mm is termed as severe, gross or frank lateral ventriculomegaly. Severe lateral ventriculomegaly is classified as:

1. Obstructive hydrocephalus due to obstruction to flow of CSF in its pathway. It is termed noncommunicating or communicating depending on whether the site of obstruction is within or outside the ventricular system.
2. Severe lateral ventriculomegaly associated with other intracranial or extracranial malformations such as malformations of cortical development (Fig. 7.5). This is described in the chapter on malformations of cortical development.

7.2.1 Obstructive Hydrocephalus

CSF flow can be obstructed at the foramen of Monro, aqueduct or fourth ventricle. The commonest site is aqueductal obstruction or stenosis (Figs. 7.6 and 7.7). The other common causes of obstruction are Chiari II and Dandy-Walker malformations (Fig. 7.8 a–c). Tumor, cyst and intraventricular hemorrhage are also causes of obstruction. Upstream dilatation of the ventricular system is seen. For example, in aqueductal stenosis, the lateral and third ventricles are dilated, whereas the fourth ventricle is not. Aqueductal stenosis can be X-linked recessive or multifactorial. The X-linked recessive form (L1CAM gene mutation) affects males and is associated with adducted thumbs. Recurring hydrocephalus in male fetuses is an indication for prenatal invasive testing for

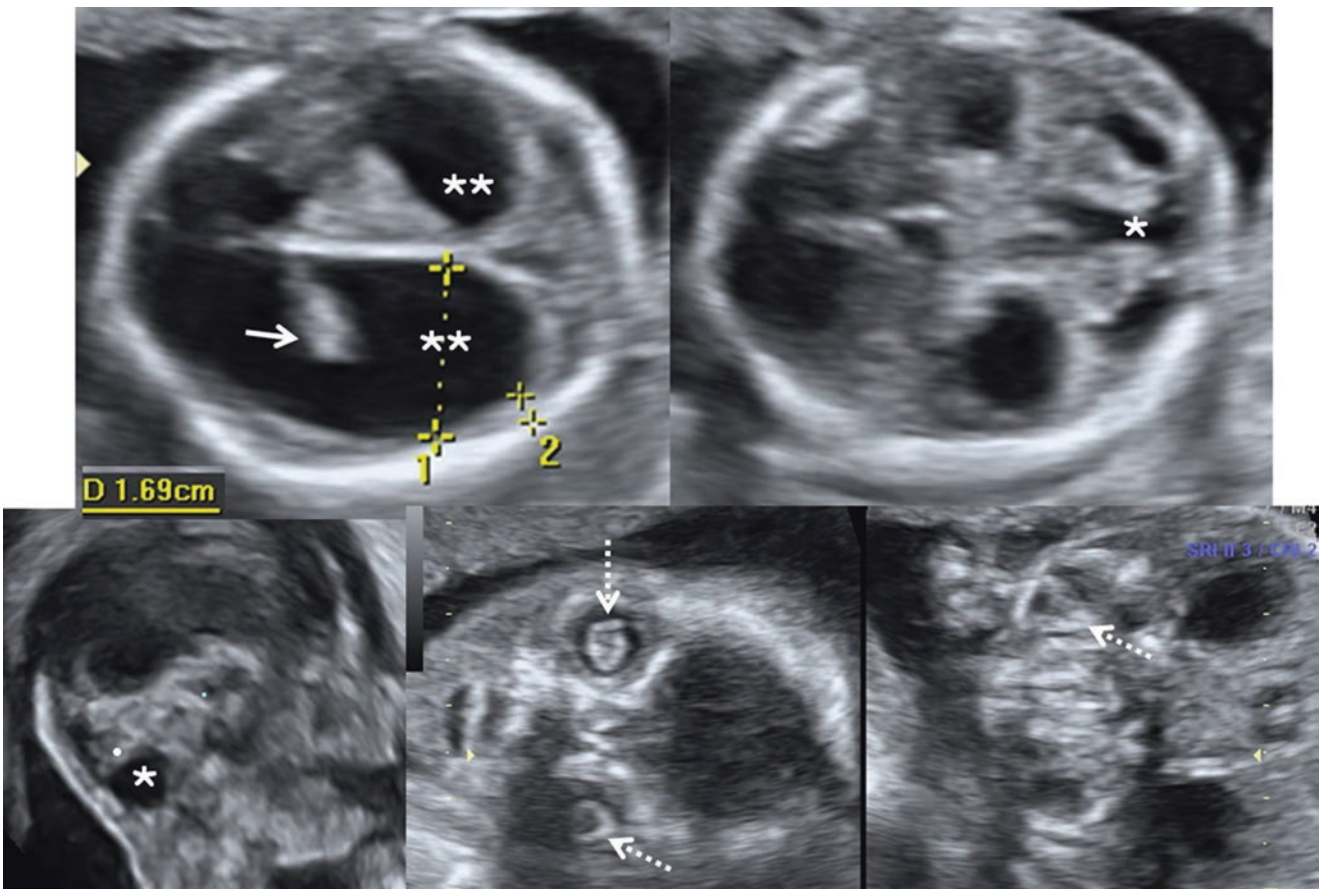


Fig. 7.5 20 weeks (TAS) *hydrocephalus, Dandy-Walker malformation and retinal dysplasia – Walker-Warburg syndrome* – axial transventricular and transcerebellar and midsagittal sections of the cranium and coronal and lateral axial sections of the orbits. Bilateral severe lateral ventriculomegaly (**), dangling choroid plexus (solid arrow), open fourth ventricle (*), rotated and elevated vermis (dot), bilateral thick dysplastic non-attached retina (dotted arrows)

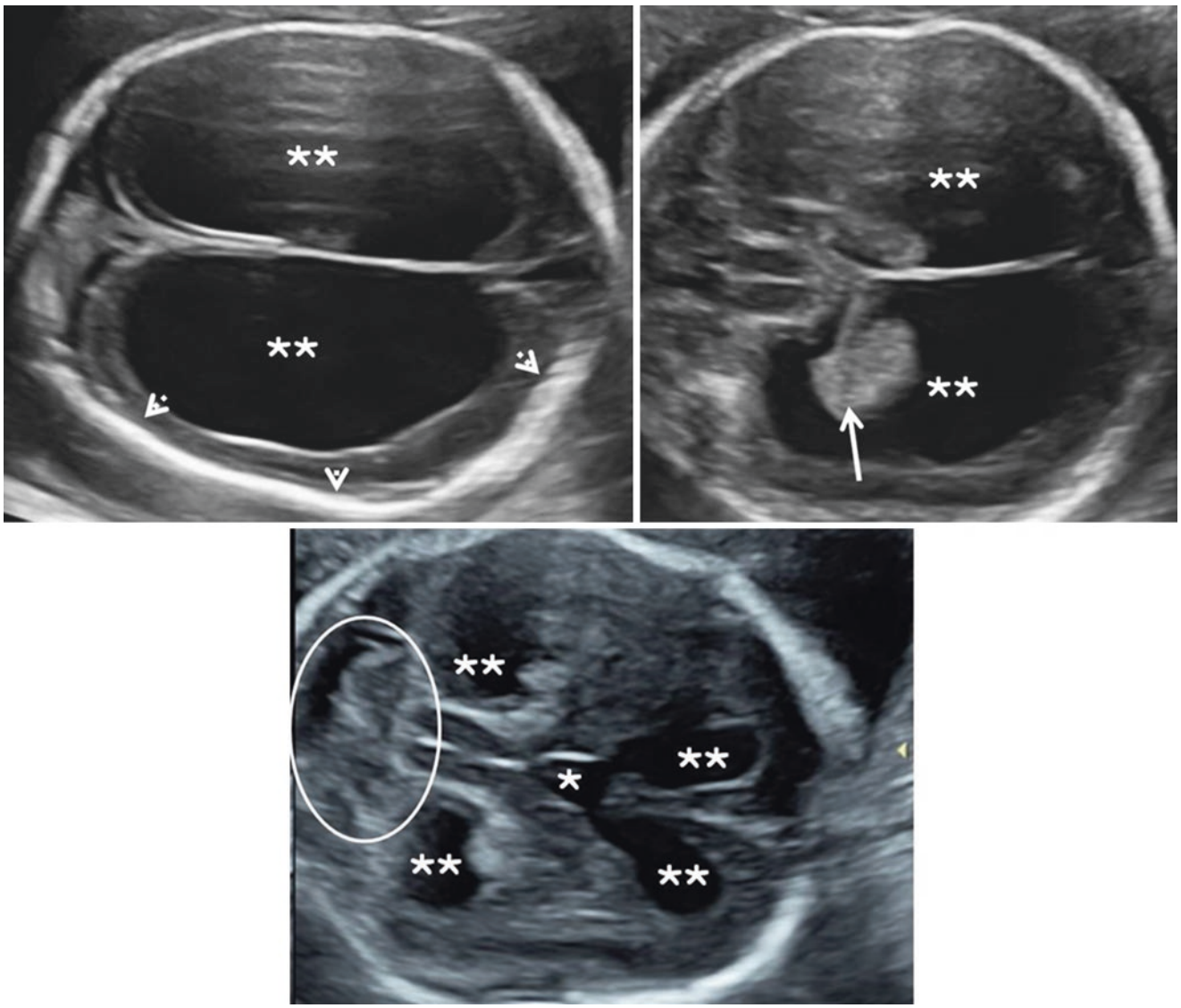


Fig. 7.6 21 weeks (TAS) *hydrocephalus due to aqueductal stenosis* – axial transventricular and transcerebellar sections – bilateral severe lateral ventriculomegaly (**), dangling choroid plexus (solid arrow), obliteration of subarachnoid space (arrowheads), dilated third ventricle (*), normal posterior cranial fossa (circled)

L1CAM gene mutation. Recurrence risk is 50% of all male progeny. The recurrence risk is 4% when the cause is multifactorial.

Intrauterine infection with CMV or toxoplasma can also cause aqueductal stenosis.

The ultrasound findings in hydrocephalus due to aqueductal stenosis are as follows:

(a) Large head circumference
(b) Cerebral parenchymal thinning
(c) Obliteration of subarachnoid space
(d) Effacement of sulci
(e) Compression, obliteration or barotraumatic destruction of the CSP
(f) Dangling choroid plexus (hinged at the foramen of Monro)
(g) Dilatation of the third ventricle and its suprapineal recess
(h) Normal posterior cranial fossa

It is important to note that obstructive hydrocephalus can manifest late in pregnancy. Hence, a normal midtrimester anomaly scan cannot rule out late-onset hydrocephalus (Fig. 7.9).

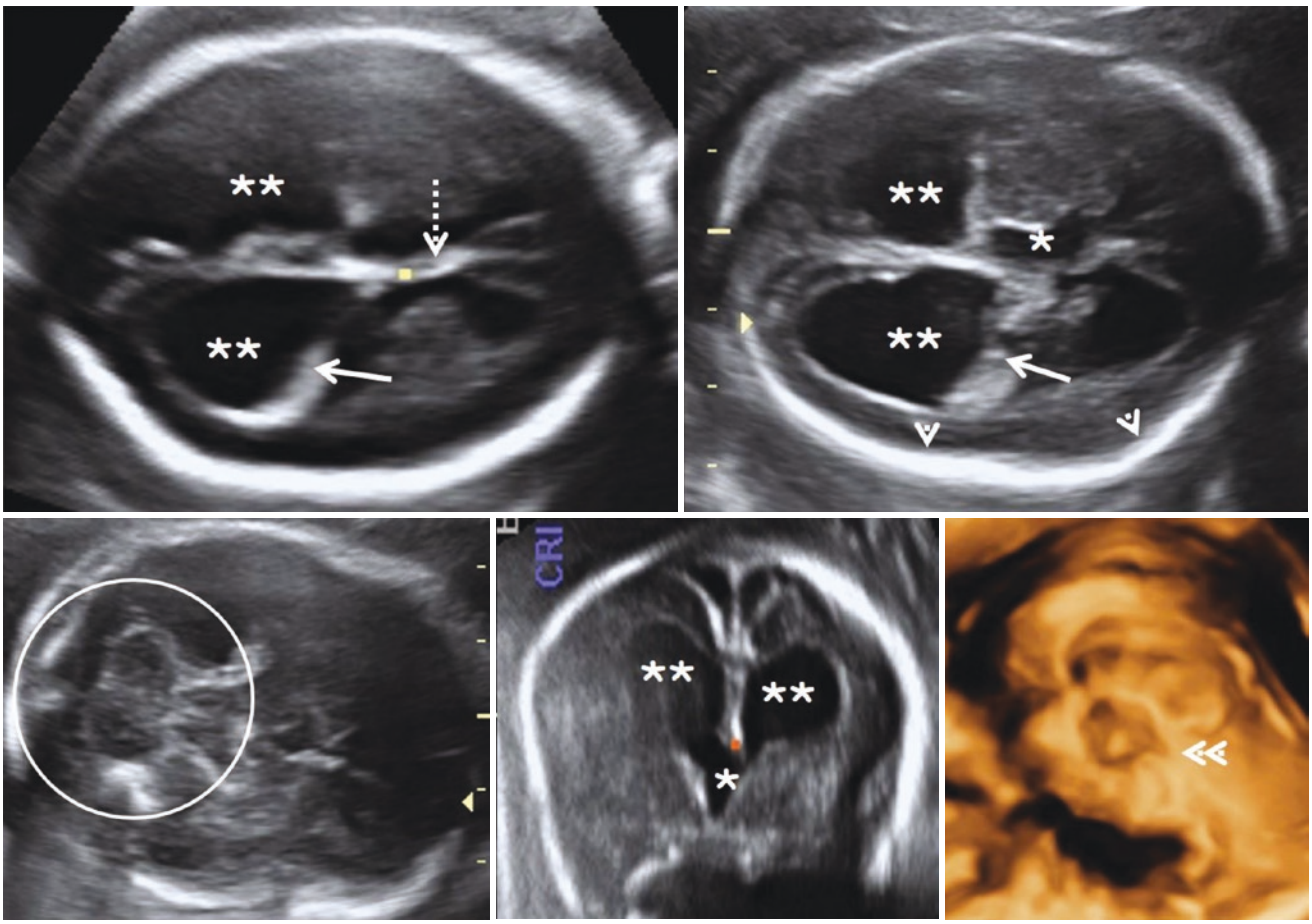


Fig. 7.7 23 weeks (TAS and 3D US) *hydrocephalus due to aqueductal stenosis* – axial transventricular, transthalamic, transcerebellar, coronal transcaudate and 3D rendered midsagittal sections – bilateral severe lateral ventriculomegaly (**), dangling choroid plexus (solid arrow), dilated third ventricle (*), obliterated subarachnoid space (arrowhead), CSP compressed with the layers apposed (dotted arrow), normal posterior cranial fossa (circled), dilated suprapineal recess (double arrowheads)

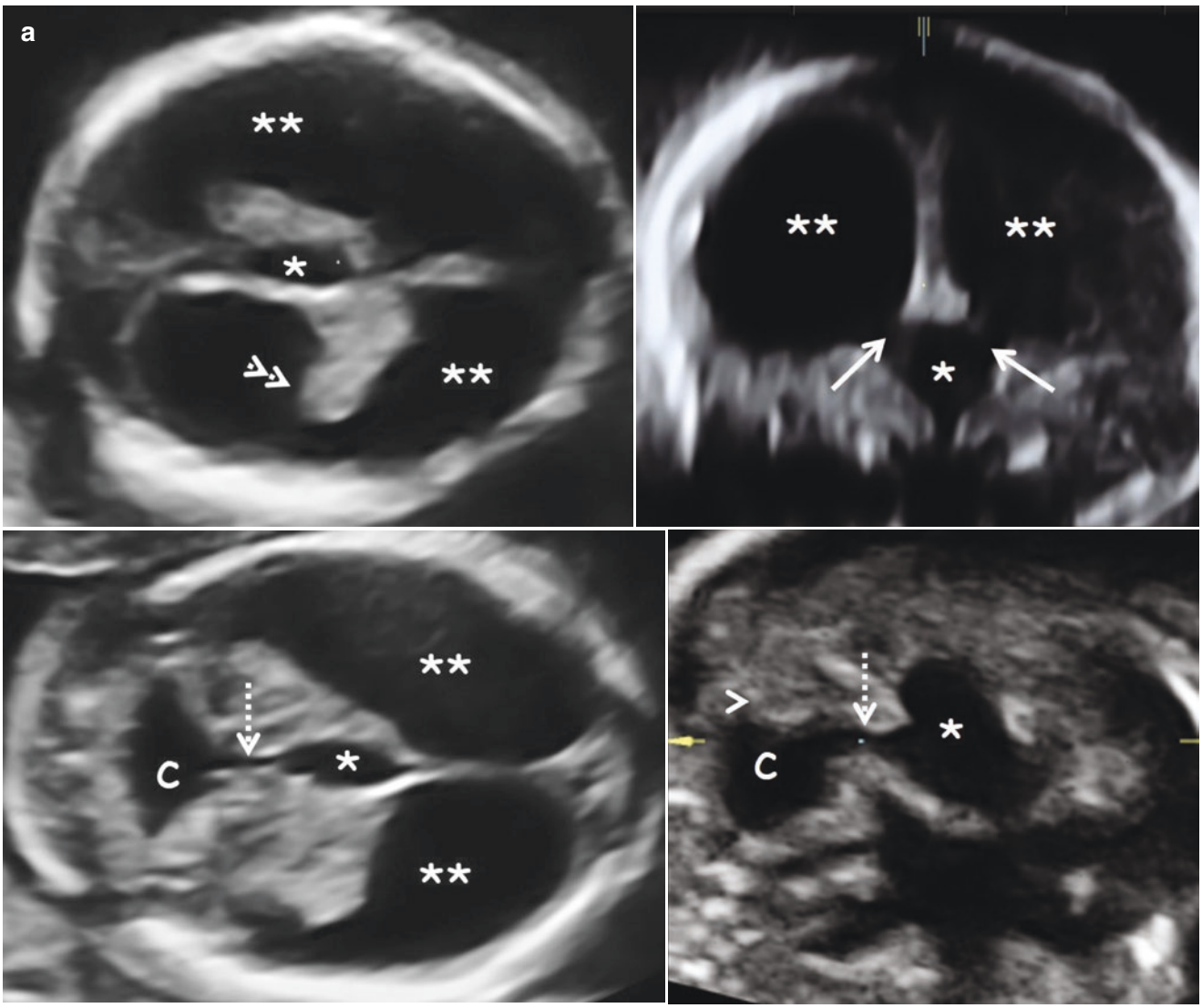
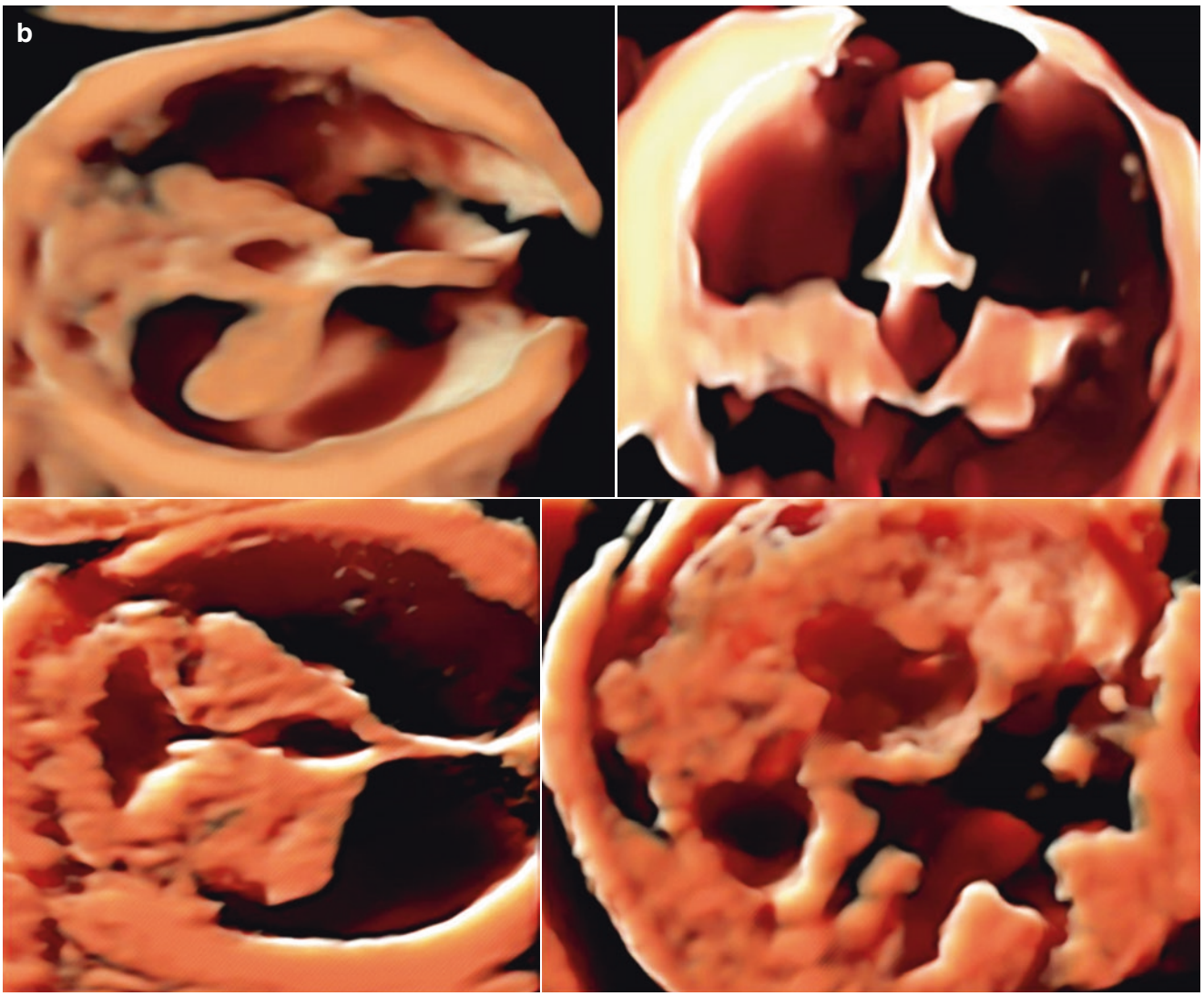


Fig. 7.8 (**a**) 18 weeks (TAS and 3D US) *hydrocephalus due to Dandy-Walker malformation with limb joint contractures* – axial transventricular, coronal transcaudate, axial transcerebellar and 3D multiplanar midsagittal sections – bilateral severe lateral ventriculomegaly (**), dangling choroid plexus (double arrowheads), posterior cranial fossa cyst (c), foramina of Monro (solid arrows), dilated aqueduct (dotted arrow), flattened and rotated vermis (arrowhead). (**b**) 18 weeks (TAS and 3D US) *hydrocephalus due to Dandy-Walker malformation with limb joint contractures* – 3D rendered axial transventricular, coronal transcaudate, axial transcerebellar and midsagittal sections – all findings as in the previous figure. (**c**) 18 weeks (TAS and 3D US) *hydrocephalus due to Dandy-Walker malformation with limb joint contractures* – coronal and sagittal sections of the limbs – bilateral elbow contractures (dotted arrow) and genu recurvatum (solid arrow). Note the limb soft tissue paucity



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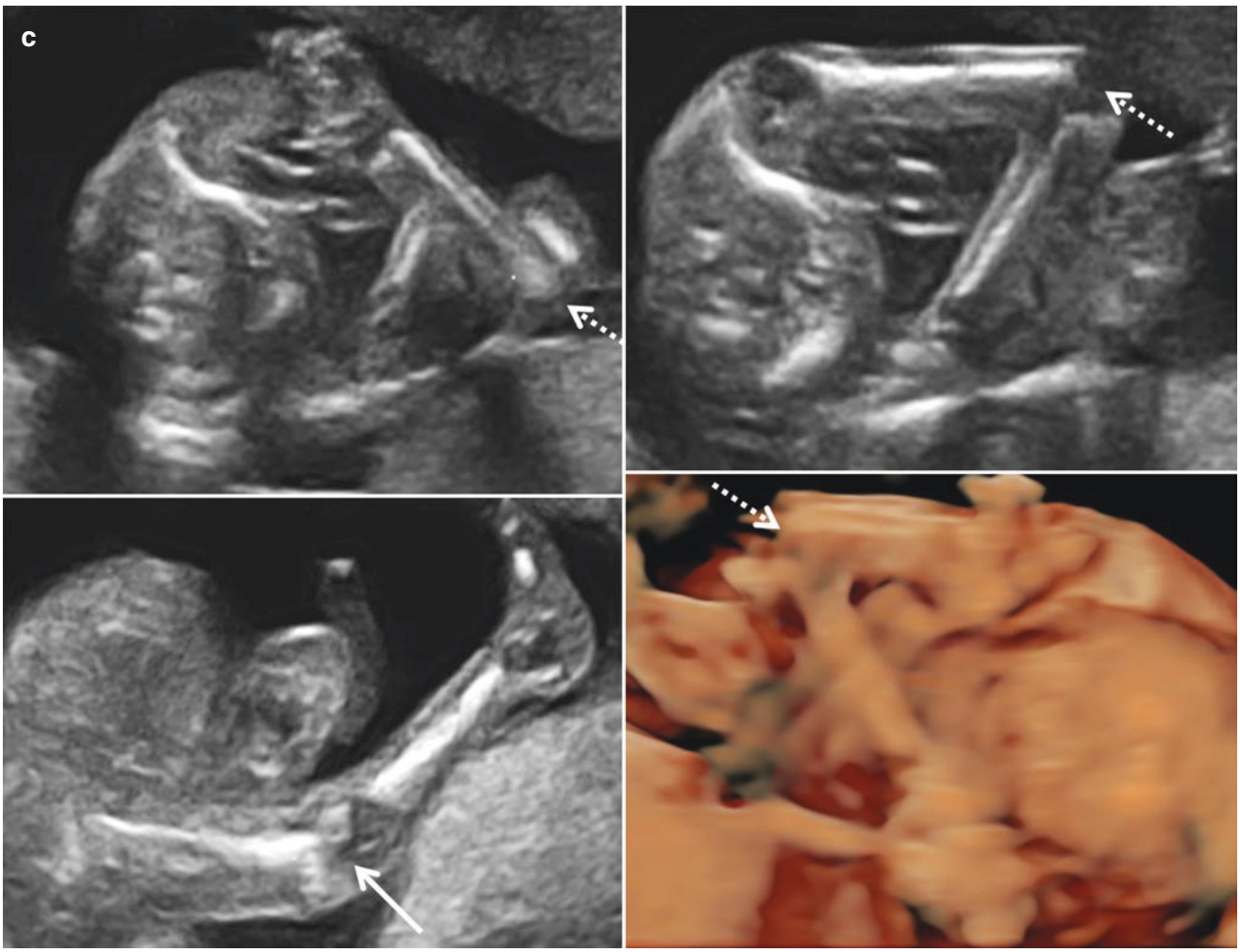

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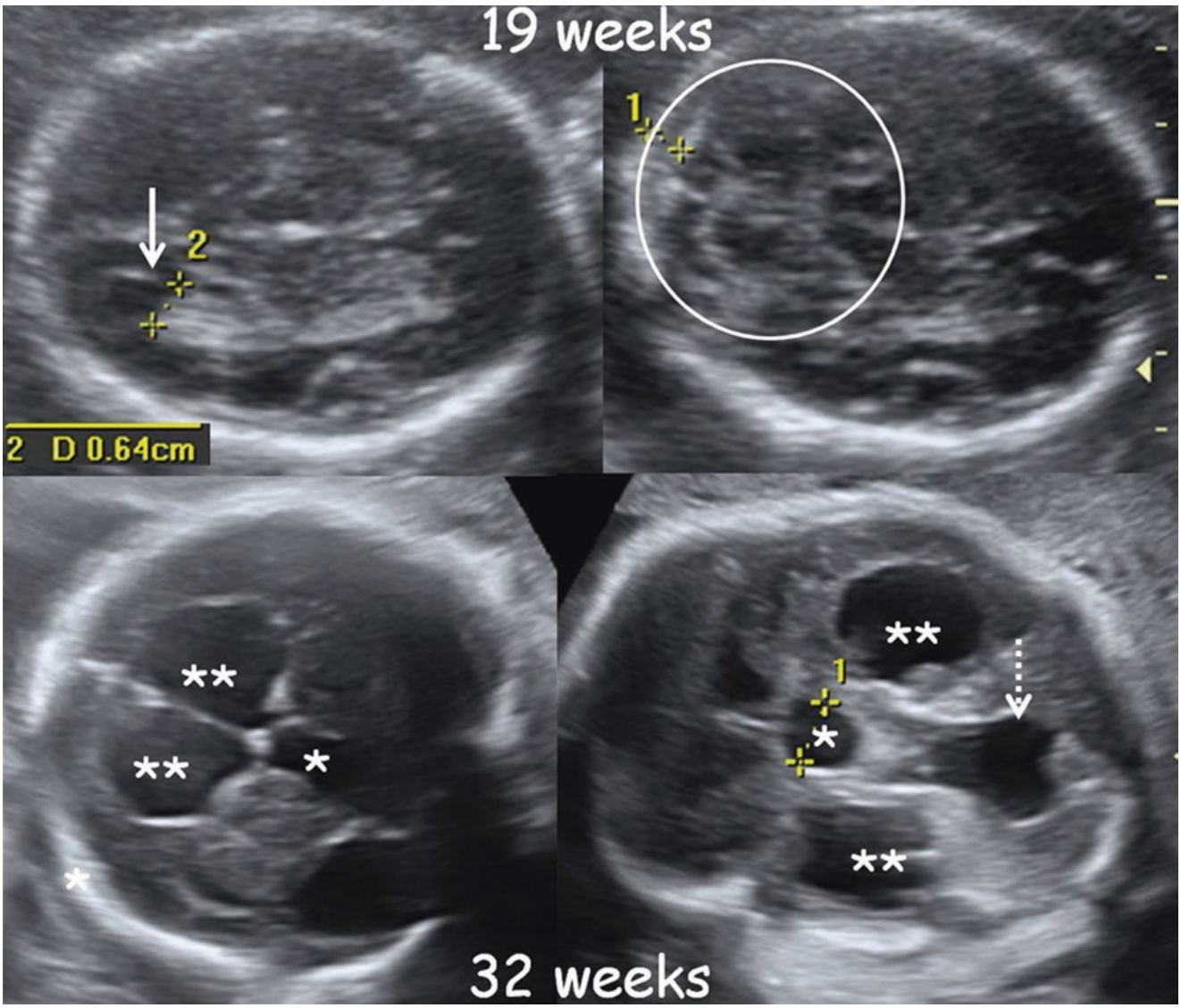

Fig. 7.9 19 and 32 weeks (TAS) *late-onset hydrocephalus (quadriventricular dilatation)* possibly due to obstruction at the foramen of Magendie – axial transventricular and transcerebellar sections at 19 weeks, coronal transthalamic and axial transcerebellar sections at 32 weeks – normal lateral ventricle (solid arrow) and posterior cranial fossa (circled) at 19 weeks, bilateral severe lateral ventriculomegaly (**), third ventriculomegaly (*) and fourth ventriculomegaly (dotted arrow) at 32 weeks

Suggested Reading

1. Melchiorre K, Bhide A, Gika AD, Pilu G, Papageorghiou AT. Counseling in isolated mild fetal ventriculomegaly. Ultrasound Obstet Gynecol. 2009;34:212–24.
2. Scala C, Familiari A, Pinas A, Papageorghiou AT, Bhide A, Thilaganathan B, Khalil A. Perinatal and long-term outcomes in fetuses diagnosed with isolated unilateral ventriculomegaly: systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2017;49:450–9.
3. Li Y, Estroff JA, Khwaja O, Mehta TS, Poussaint TY, Robson CD, Feldman HA, Ware J, Levine D. Callosal dysgenesis in fetuses with ventriculomegaly: levels of agreement between imaging modalities and postnatal outcome. Ultrasound Obstet Gynecol. 2012;40:522–9.
4. Guibaud L, Lacalm A. Etiological diagnostic tools to elucidate 'isolated' ventriculomegaly. Ultrasound Obstet Gynecol. 2015;46:1–11.