CNS Anomalies Between 11 and 14 Weeks

8

The intracranium is routinely assessed in the 11–14 weeks US examination. The intracranium is better studied during the 12–14 weeks gestational age than the 11th week. The anatomy of the brain at this stage differs from that in the mid trimester. The corpus callosum has not yet developed, and hence the cavum septum pellucidum (CSP) is not seen. The vermis is yet to develop, and hence the 'open fourth ventricle' is a normal finding. The normal sonoanatomy in the axial transventricular, transthalamopeduncular and transcerebellar sections and midsagittal section has been described in the Chap. 1.

Screening for open neural defects and diagnosis of anencephaly, iniencephaly, encephalocele and holoprosencephaly are discussed in this chapter.

8.1 Screening for Open Neural Tube Defects

The intracranial translucency (IT) has been described in the chapter on sonoanatomy. In open spina bifida (OSB), there is loss of CSF through the defect, resulting in a caudal displacement of the brainstem and the cerebellum (Chiari II malformation). In the second trimester, this is seen as the 'banana' sign. In the first trimester (11–14 weeks), the abnormal posterior fossa findings in OSB serve as screening parameters. These findings may be described as subjective and objective signs.

Subjective signs:

1. IT is obliterated (Figs. 8.1, 8.2, 8.3 and 8.4).
2. The brainstem appears thick.
3. The brainstem may be kinked (Fig. 8.4).

Objective signs:

1. The brainstem (BS) diameter is increased (more than 95th percentile) (Figs. 8.1, 8.2, 8.3 and 8.4).
2. The brainstem occipital bone distance (BSOB) is decreased (less than 5th percentile).
3. The BS to BSOB ratio is increased (more than 95th percentile).

These findings are not present in closed spina bifida.

When there are positive findings on screening, one must examine the fetus for an open spina bifida or an encephalocele (Figs. 8.5 and 8.6). This is best done by the transvaginal approach. If the transvaginal examination is normal, an US examination at 15 weeks should be advised. It should be emphasised that the diagnosis of OSB is made only if the actual defect is seen. The osseous and soft tissue findings in OSB are described in the chapter on anomalies of dorsal induction.

In a recent meta-analysis and systematic review, the sensitivity and specificity of IT in the detection of open spina bifida were 53.5% and 99.7%, respectively.

Other findings in open spina bifida:

1. BPD may be lesser than normal.
2. Parallelism of the thalami and cerebral peduncles (parallel peduncle sign) is seen. The midbrain is juxtaposed to the occipital bone. The distance between the aqueduct and the occipital bone is decreased (Figs. 8.2 and 8.3).
3. Occasionally the 'lemon' and 'banana' signs are present as early as 13–14 weeks (Figs. 8.2 and 8.3).
4. Kyphoscoliosis may be seen. Hydrocephalus and lower limb neuromuscular sequelae usually appear in the second or third trimesters.

An increased IT and BSOB distance (more than 95th percentile) and decreased BS to BSOB ratio (less than 5th percentile) are associated with cystic lesions of the posterior cranial fossa such as Blake's pouch cyst, vermian hypoplasia and Dandy-Walker malformation (Fig. 8.7). These lesions are detectable after 20 weeks. It is, therefore, important to carefully examine the posterior fossa in the second trimester in cases of increased IT.

B. S. Rama Murthy, *Imaging of Fetal Brain and Spine*, https://doi.org/10.1007/978-981-13-5844-9_8

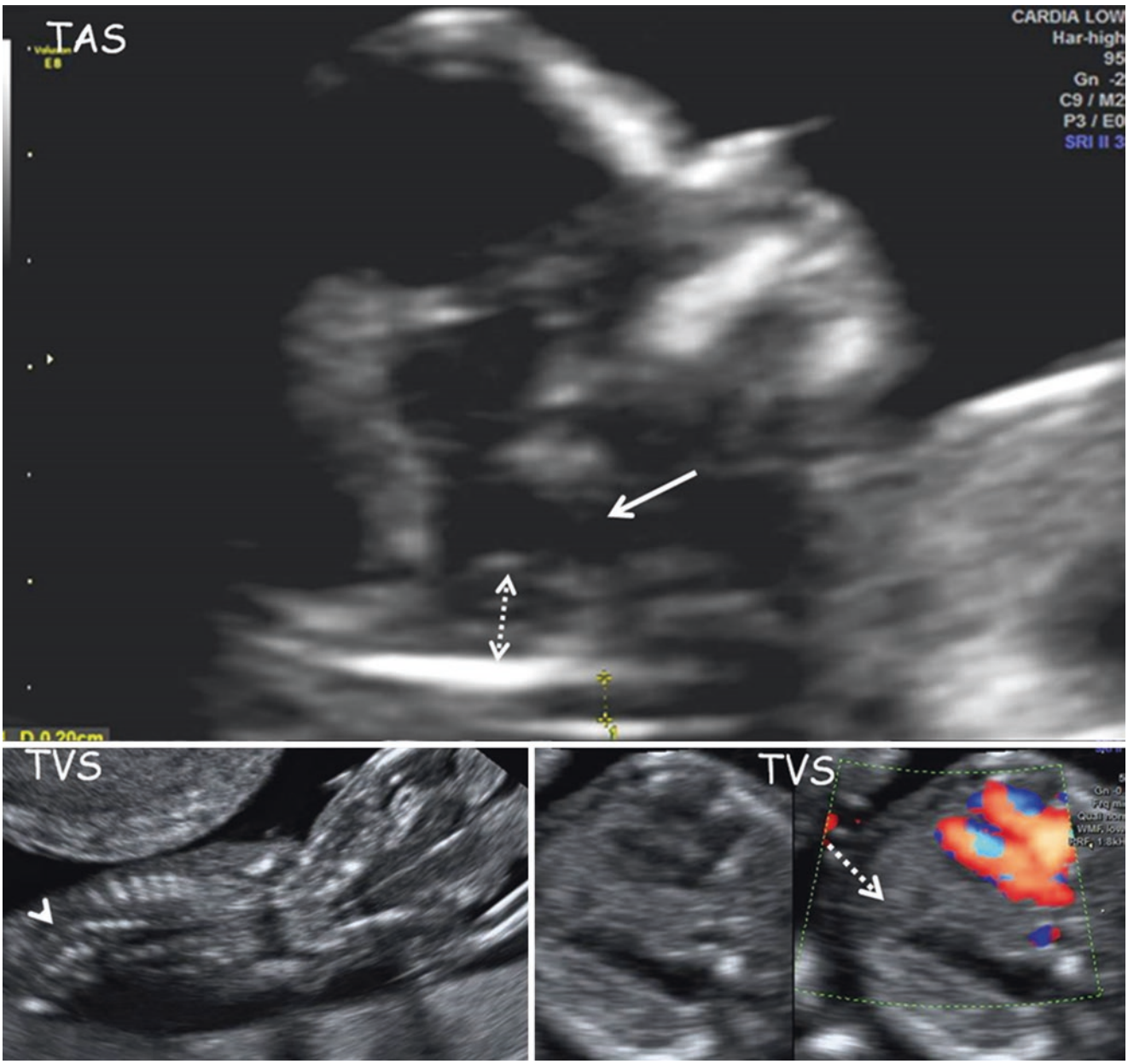

Fig. 8.1 13 weeks (TAS and TVS) *abnormal IT – lumbosacral open spina bifida* – midsagittal section, TVS coronal section of spine, four-chamber view B mode and color Doppler. IT is obliterated, the brainstem (solid arrow) is thick, brainstem diameter is more than brainstem occipital bone distance (double-headed dotted arrow), splaying of the posterior ossific centres indicates lumbosacral open spina bifida (arrowhead), associated finding of left congenital diaphragmatic hernia (dotted arrow)

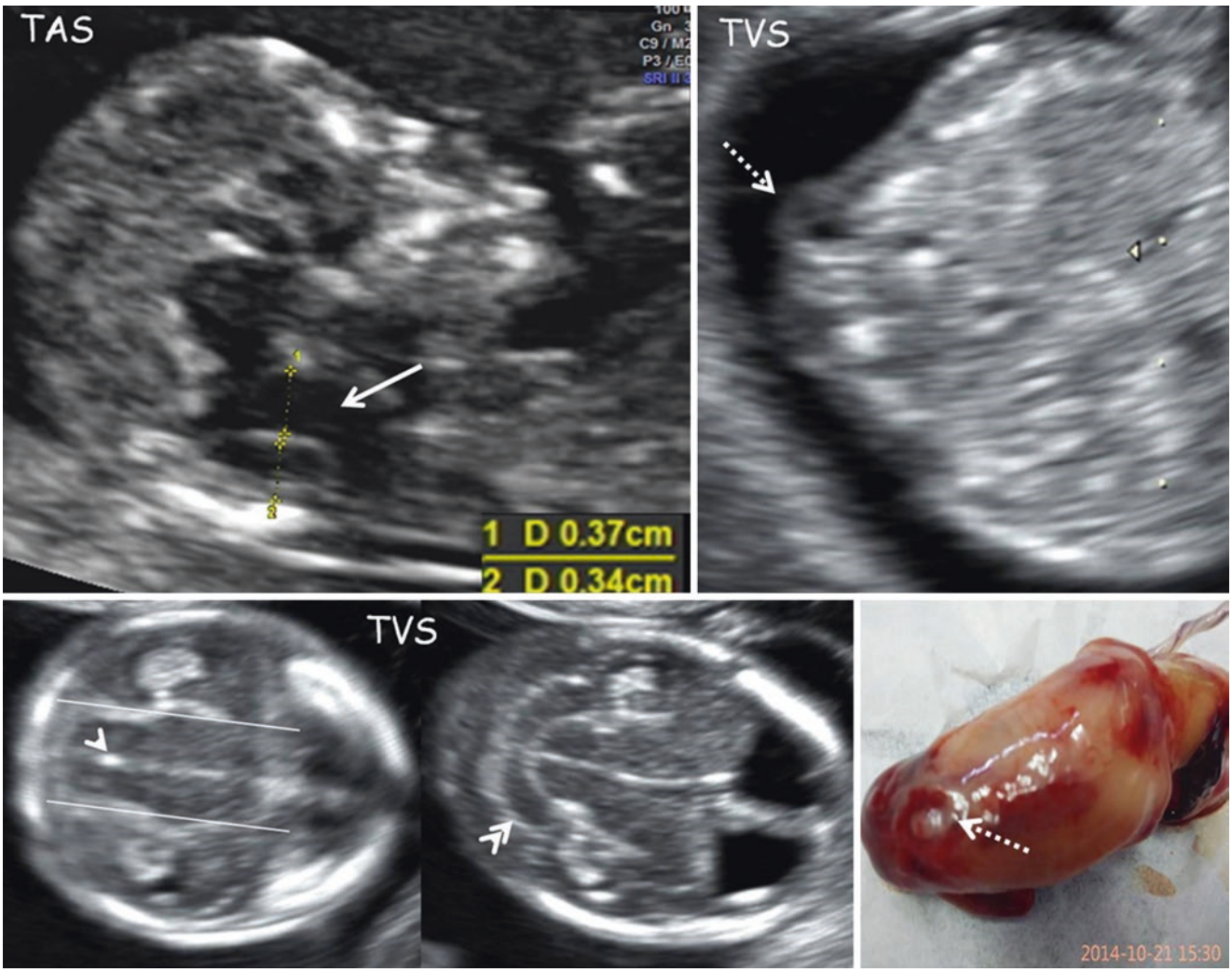

Fig. 8.2 13 weeks (TAS and TVS) *abnormal IT – lumbar open spina bifida* – midsagittal section, axial section of lumbar spine, axial thalamopeduncular and transcerebellar sections of the cranium, picture of abortus - IT is obliterated, the brainstem (solid arrow) is thick, brainstem diameter is more than brainstem occipital bone distance, lumbar myelomeningocele (dotted arrow), parallel peduncle sign (straight lines), aqueduct close to the occipital bone (arrowhead), 'banana' sign (double arrowhead)

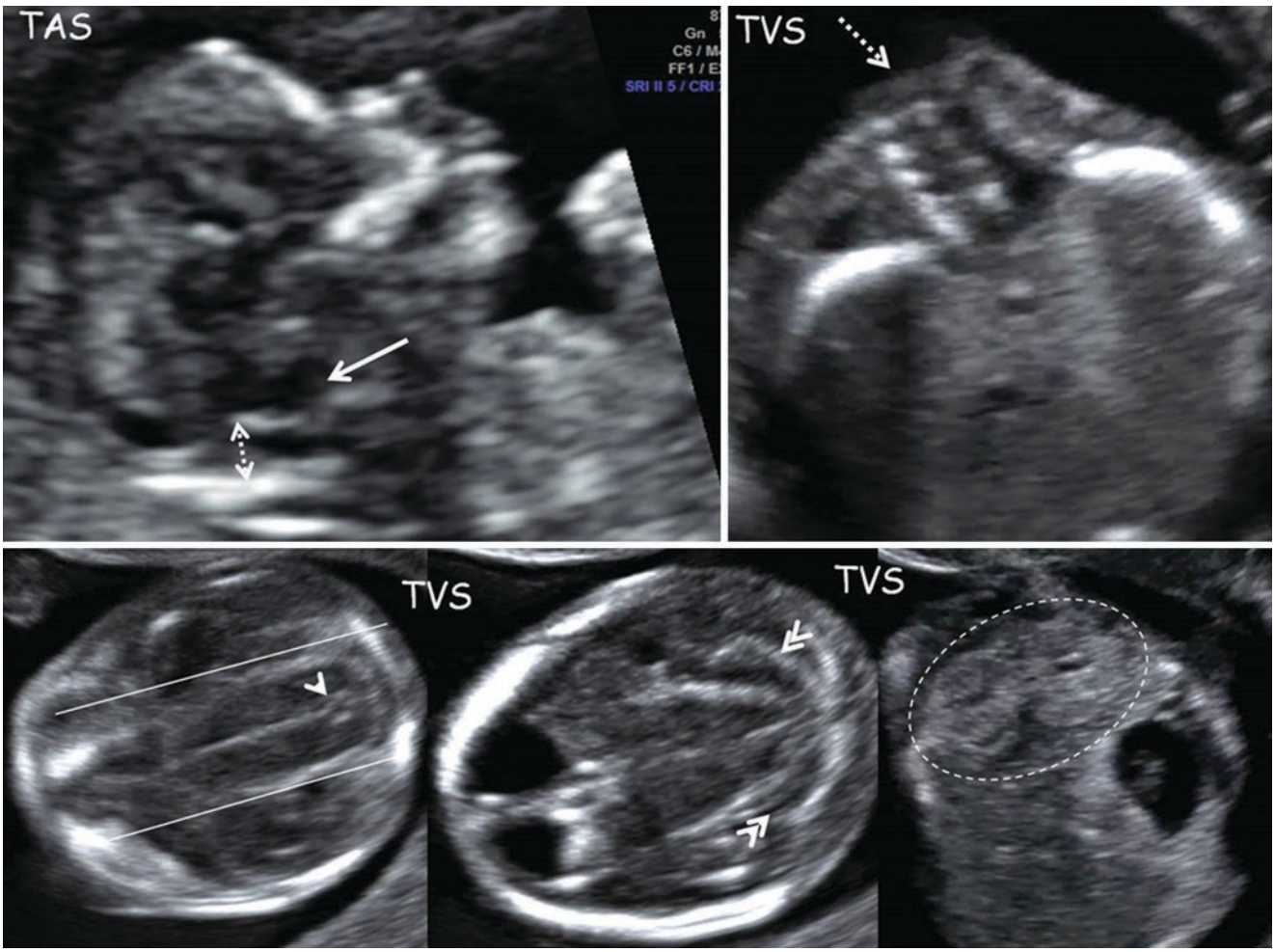

Fig. 8.3 13 weeks (TAS and TVS) *abnormal IT – lumbar open spina bifida* – midsagittal section, oblique coronal section of lumbar spine, axial transthalamopeduncular and transcerebellar sections, axial section of the upper abdomen - IT is obliterated, the brainstem (solid arrow) is thick, brainstem diameter is more than the brainstem occipital bone distance (double-headed dotted arrow), lumbar open spina bifida (dotted arrow), parallel peduncle sign (straight lines), aqueduct close to the occipital bone (arrowhead) and the 'banana' sign (double arrowhead), kidneys on either side are fused across the midline (horseshoe kidney) (circled)

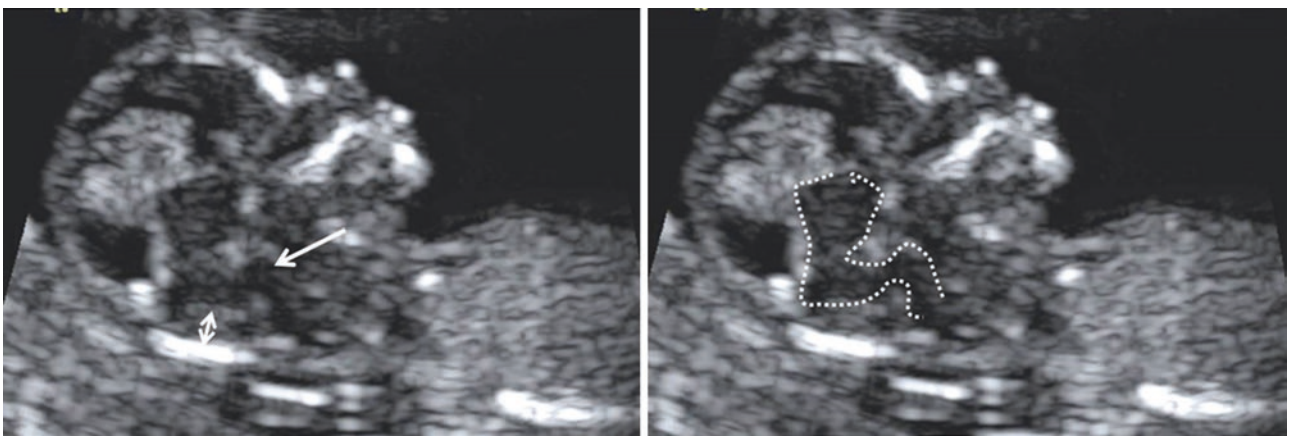

Fig. 8.4 12 weeks (TAS) *abnormal IT, kinked, thick brainstem* – IT is obliterated, the brainstem (solid arrow) is thick and kinked, brainstem diameter is more than the brainstem occipital bone distance (double-headed arrow). This fetus had a lumbar open spina bifida

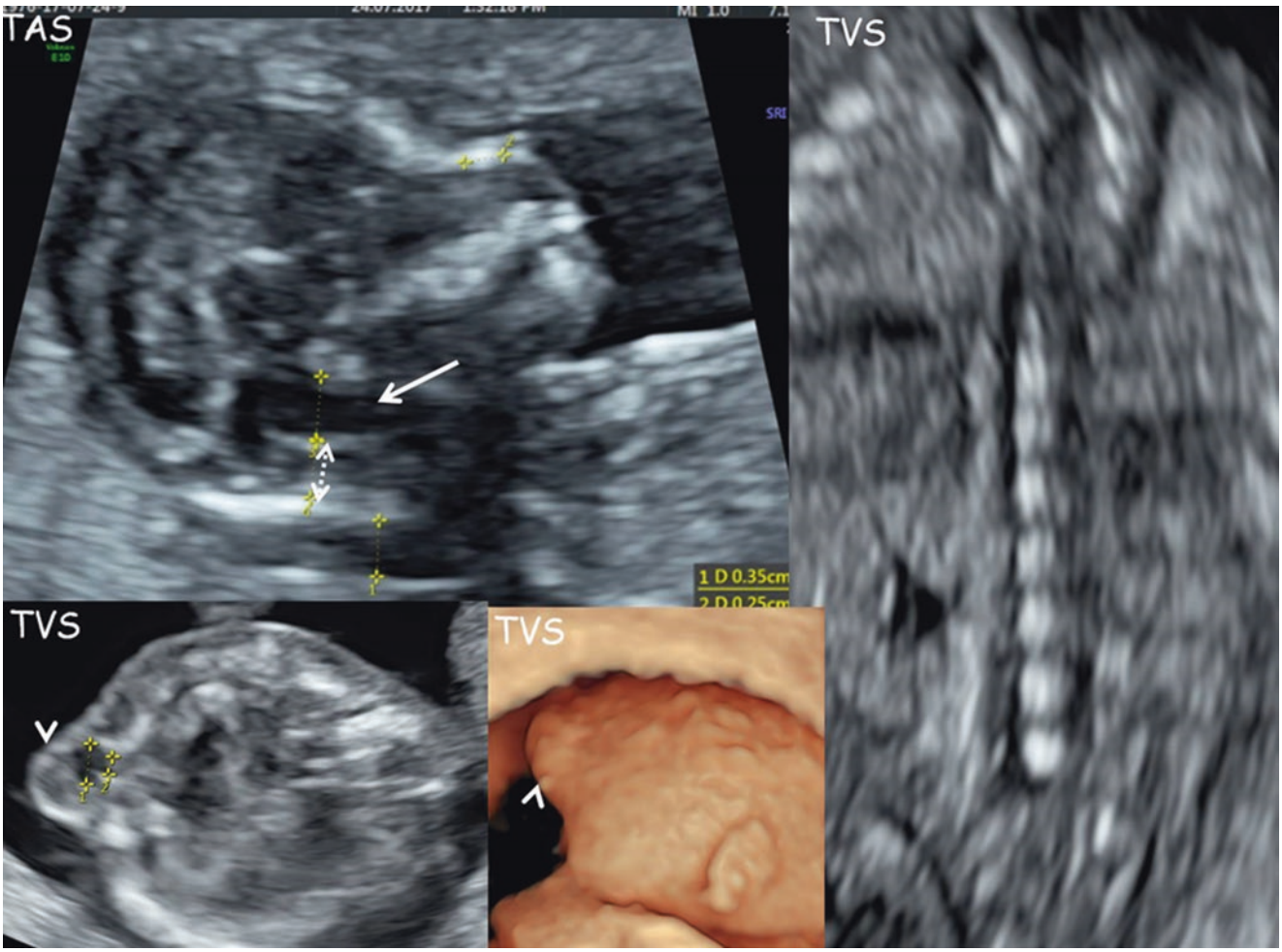

Fig. 8.5 13 weeks (TAS, TVS and 3D US) *abnormal IT – occipital encephalocele* – midsagittal section, coronal section of thoracolumbar spine, axial transventricular section, 3D surface rendering of fetal cranium - IT is obliterated, the brainstem (solid arrow) is thick, the brainstem diameter is more than the brainstem occipital bone distance (double-headed dotted arrow), fetal spine is normal, small occipital encephalocele (arrowhead)

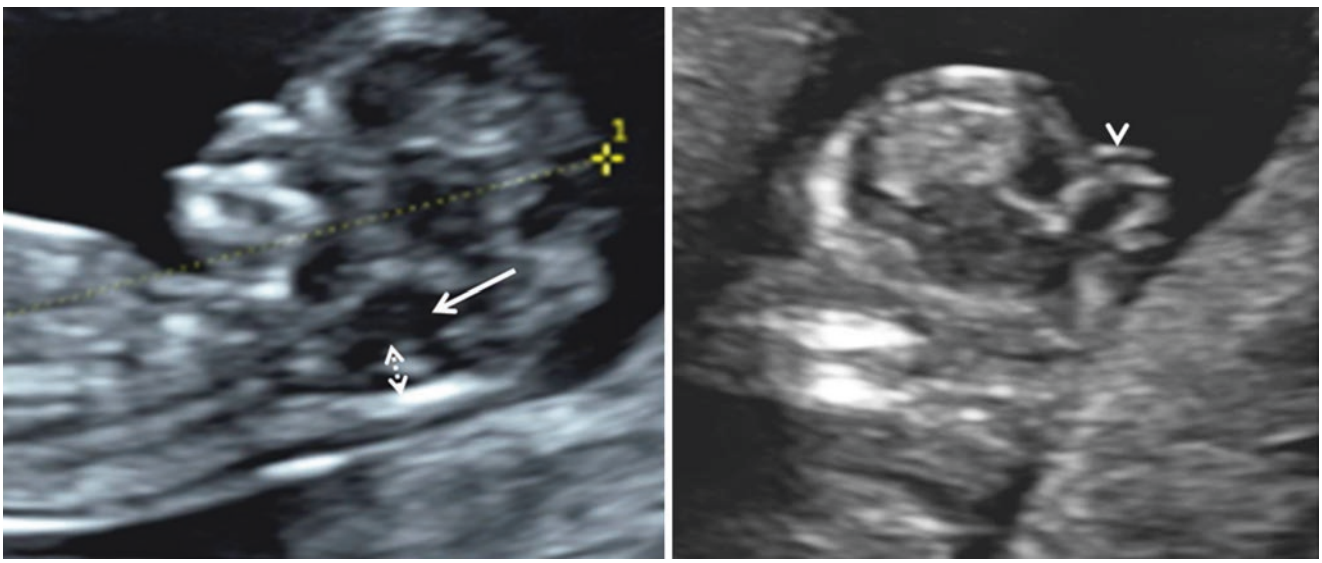

Fig. 8.6 12 weeks (TAS) *abnormal IT – occipital encephalocele* – midsagittal section, oblique sagittal section of the cranium - IT is obliterated, the brainstem (solid arrow) is thick, the brainstem diameter is more than the brainstem occipital bone distance (double-headed dotted arrow), very small occipital encephalocele (arrowhead), fetal spine was normal

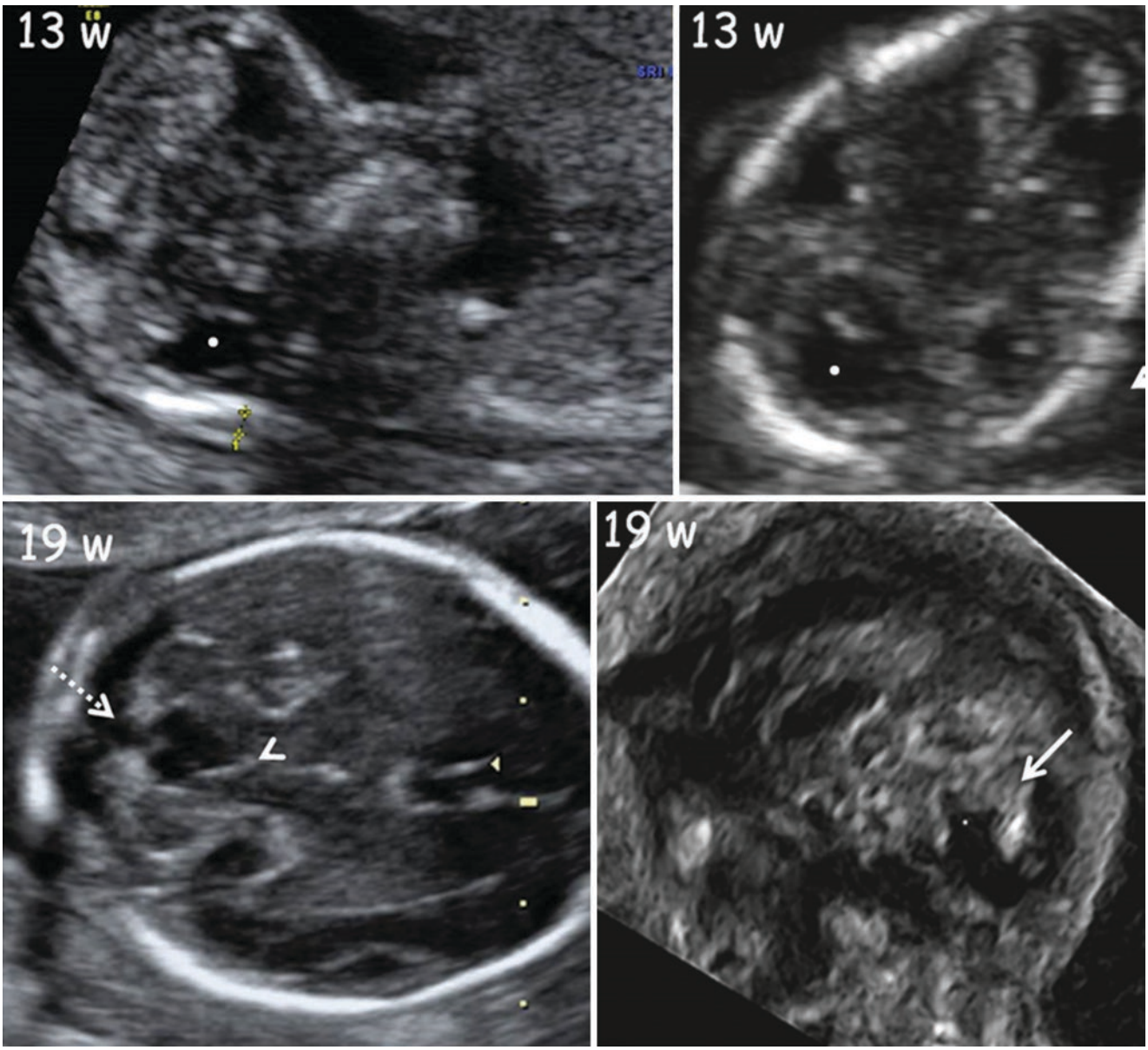

Fig. 8.7 Third-degree consanguineous couple with an 11-year-old child with *Joubert syndrome* – 13 and 19 weeks (TAS) midsagittal and axial transcerebellar sections at 13 weeks, axial transcerebellar and midsagittal sections at 19 weeks – *increased intracranial translucency* (dot), normal brainstem, BSOB distance is increased, 'open fourth ventricle' (dotted arrow), the floor of the fourth ventricle is pointed anteriorly (arrowhead), the vermis is small and rotated (solid arrow). Findings indicate recurrence of Joubert syndrome. First clue was an increased IT in the 13 weeks examination

8.2 Exencephaly-Anencephaly Sequence

Failure of closure of the rostral neuropore on the day 24 of conceptual age results in the lethal exencephaly-anencephaly sequence. Exencephaly is the presence of the forebrain with the absence of the calvarium and skin. A complete absence of the forebrain, calvarium and skin is anencephaly. With time, exencephaly evolves to anencephaly and hence is termed a sequence.

The ultrasound findings are as follows:

1. The echogenic, ossified calvarial bones are not seen (Fig. 8.8).
2. The brain tissue is seen exposed to amniotic fluid.
3. The midline cranial (falx) echoes are not seen. The 'butterfly' sign is absent.
4. The brain appears disorganised and irregular in contour.
5. Eventually the brain is not seen (resorbed) resulting in anencephaly.

6. Loose spongy appearing tissue may be seen floating in the region of the brain.
7. The facial profile is abnormal. The forehead and rounded contour of the crown are absent.
8. The base of the skull and the orbits are present. Prominent orbits result in 'frog facies' (Fig. 8.9).
9. The CRL is lesser than expected.
10. Calvarial disruption due to amniotic bands can result in anencephaly. Asymmetry of the residual calvarium, presence of amniotic bands and other signs of disruption such as digit or limb amputation and gastroschisis are diagnostic of amniotic band sequence. There is no increased risk of recurrence in future pregnancies.
11. OSB, rachischisis and iniencephaly may be seen with anencephaly. Cardiac, genitourinary, facial and other anomalies may be associated.

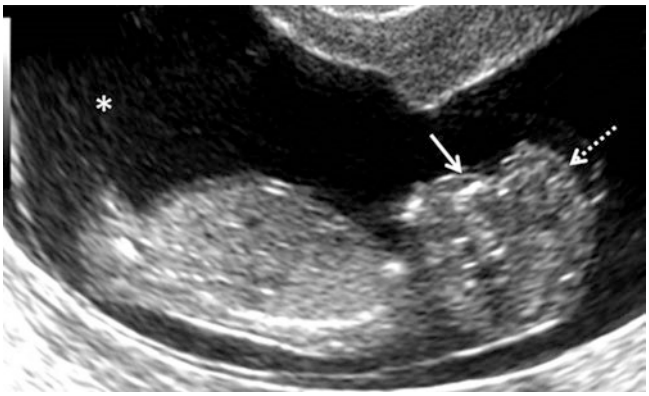

Fig. 8.8 12 weeks (TVS) *exencephaly* – sagittal section – calvarial bones not seen, brain tissue is misshapen and disorganised (dotted arrow), the face and base of skull bones are present (solid arrow), facial profile is abnormal, amniotic fluid is turbid (*)

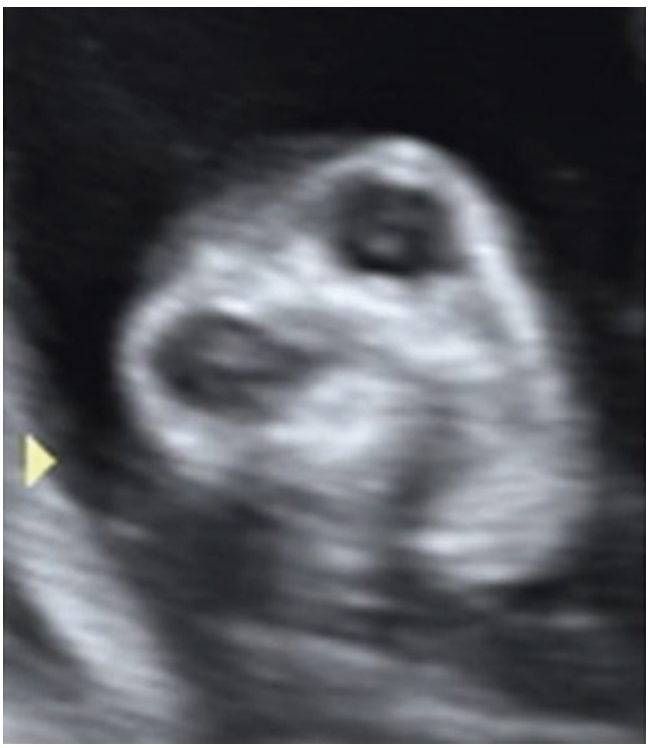

Fig. 8.9 12 weeks (TAS) coronal section – anencephaly – 'frog facies' appearance

8.3 Iniencephaly

Iniencephaly is a lethal abnormality characterised by a defect in the occipital bone, retroflexion of the spine and open spinal defect.

The ultrasound findings are as follows:

1. The head is held in persistent and severe extension. The face looks upwards with the occiput directed downwards.
2. The spine is short and lordotic with cervical and upper thoracic vertebral defects (Fig. 8.10).
3. Occipital encephalocele (protruding through the foramen magnum) may be seen.
4. The neck is absent.
5. Associated defects include anencephaly, microcephaly, hydrocephalus, open spina bifida and facial clefts.

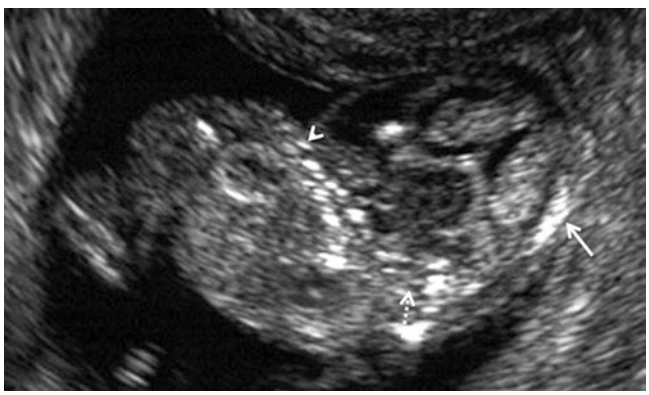

Fig. 8.10 12 weeks (TVS) *iniencephaly – sagittal section* – calvarial bones are seen (solid arrow), persistent extension at the craniovertebral junction (dotted arrow), short and disorganised spine (arrowhead)

Differential diagnosis is limb body wall complex characterised by the presence of extracorporeal viscera.

8.4 Cephalocele

A cephalocele is herniation of meninges and the brain (encephalocele) or meninges alone (meningocele) through a bony defect in the calvarium. Occipital cephaloceles are the most common. They can also occur in the frontal, parietal and basal regions. Parietal cephaloceles are due to amniotic band sequence and are disruptive in origin (Fig. 8.11a–c).

The ultrasound findings are as follows:

1. Thin-walled cystic swelling arising from the occipital (Fig. 8.12) or frontal regions (Fig. 8.13) seen on midsagittal and axial sections.
2. Presence or absence of the brain in the lesion should be assessed (Fig. 8.14).

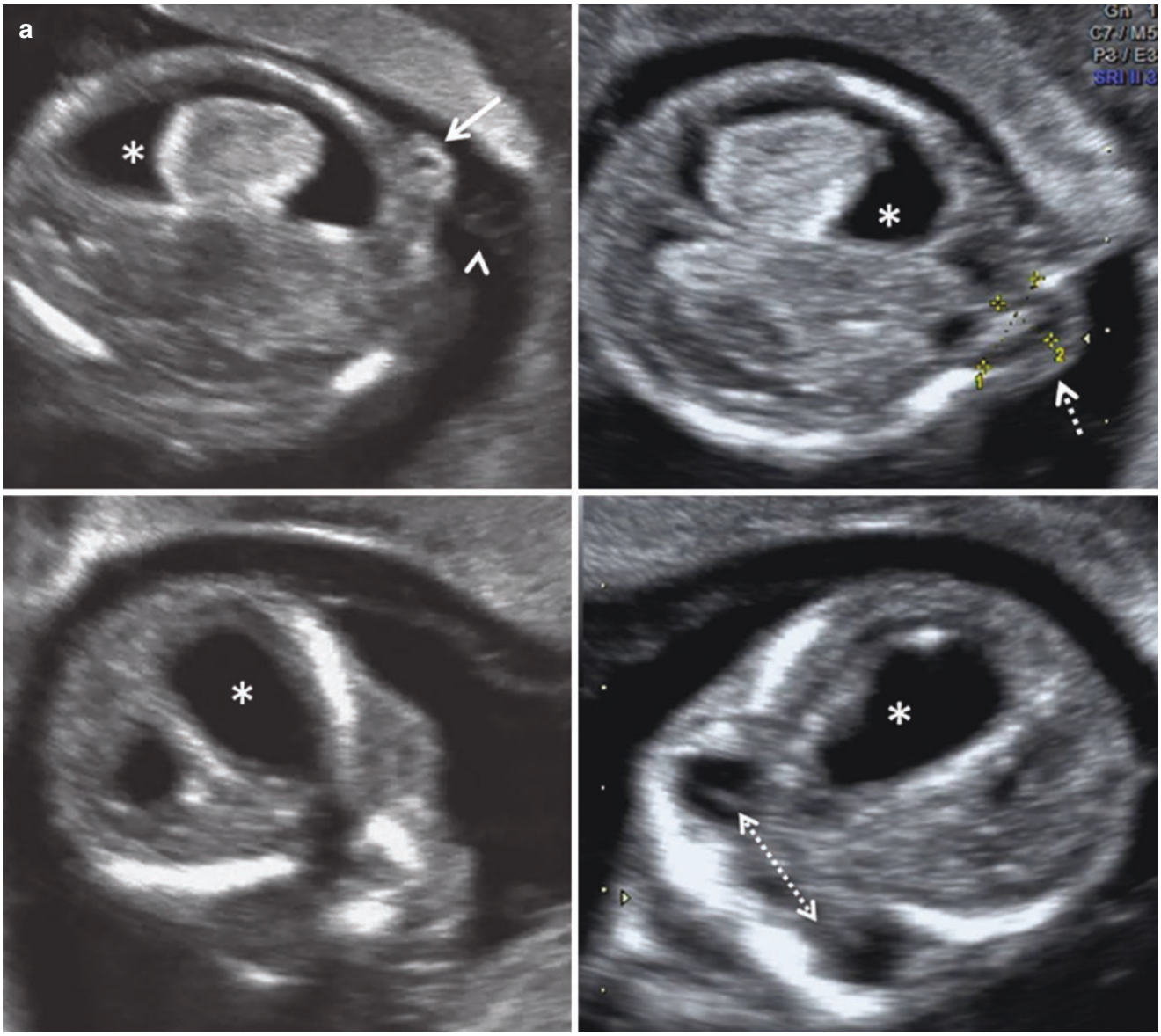

Fig. 8.11 (**a**) 13 weeks (TVS) *two encephaloceles due to amniotic band sequence* – axial transventricular and coronal transfrontal sections of the cranium, lateral axial section of the orbits – small right parieto-occipital encephalocele (solid arrow), wispy amniotic bands (arrowhead), relatively larger occipital encephalocele (dotted arrow), unilateral right lateral ventriculomegaly (*), hypertelorism (double-headed dotted arrow). (**b**) 13 weeks (TVS 3D US) *two encephaloceles due to amniotic band sequence* – oblique sections of the cranium with 3D rendered images – small right parieto-occipital encephalocele (solid arrow) with the amniotic band (arrowhead), relatively larger occipital encephalocele (dotted arrow) with the amniotic band (double arrowhead). (**c**) 13 weeks (TVS 3D US) *two encephaloceles due to amniotic band sequence* – sections of the upper and lower limbs and 3D rendered image of the upper limbs – multiple amniotic bands attached to the digits

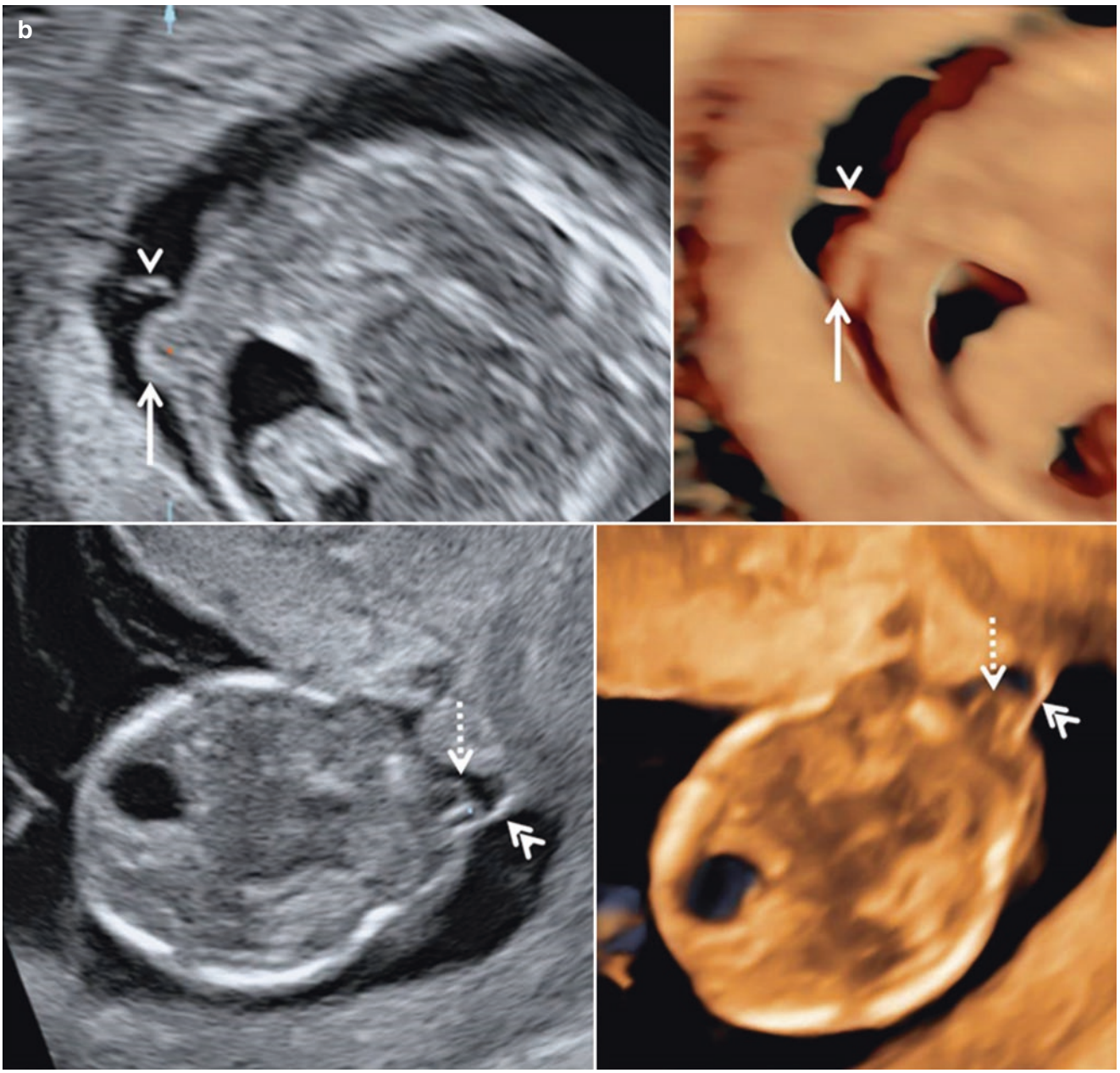

Fig. 8.11 (continued)

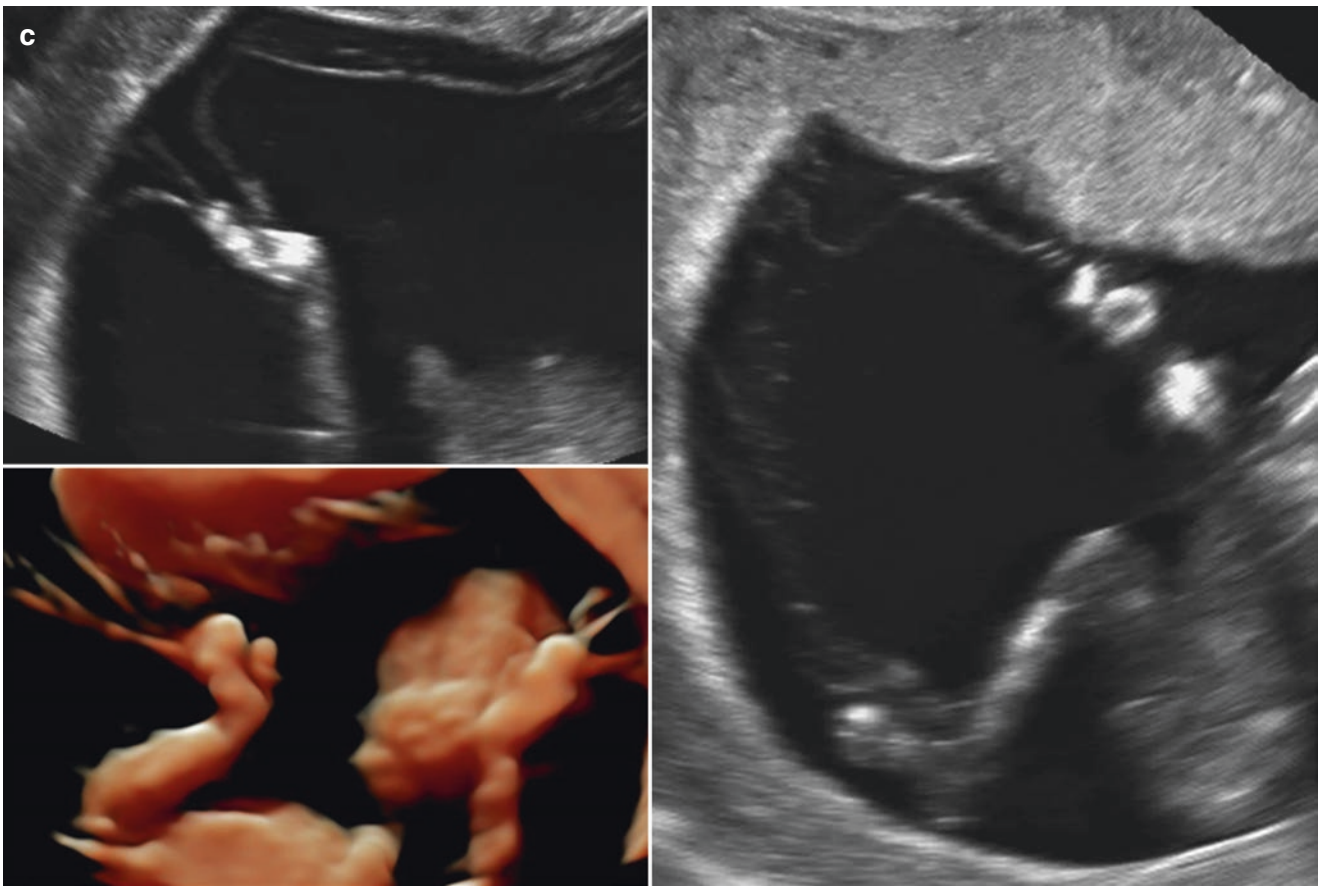

Fig. 8.11 (continued)

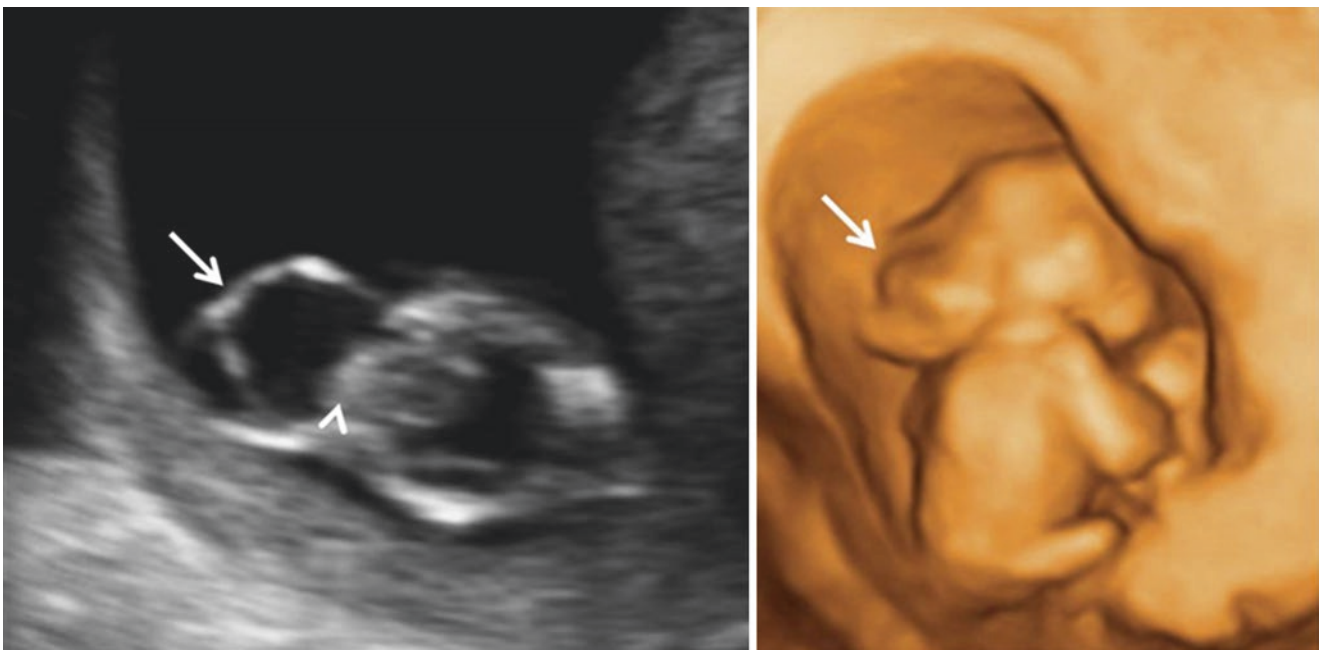

Fig. 8.12 11 weeks (TAS) *occipital encephalocele* – axial section of cranium, 3D surface rendering of the cranium – thin-walled cyst arising from the occipital region (solid arrow), brain tissue herniating into the encephalocele (arrowhead)

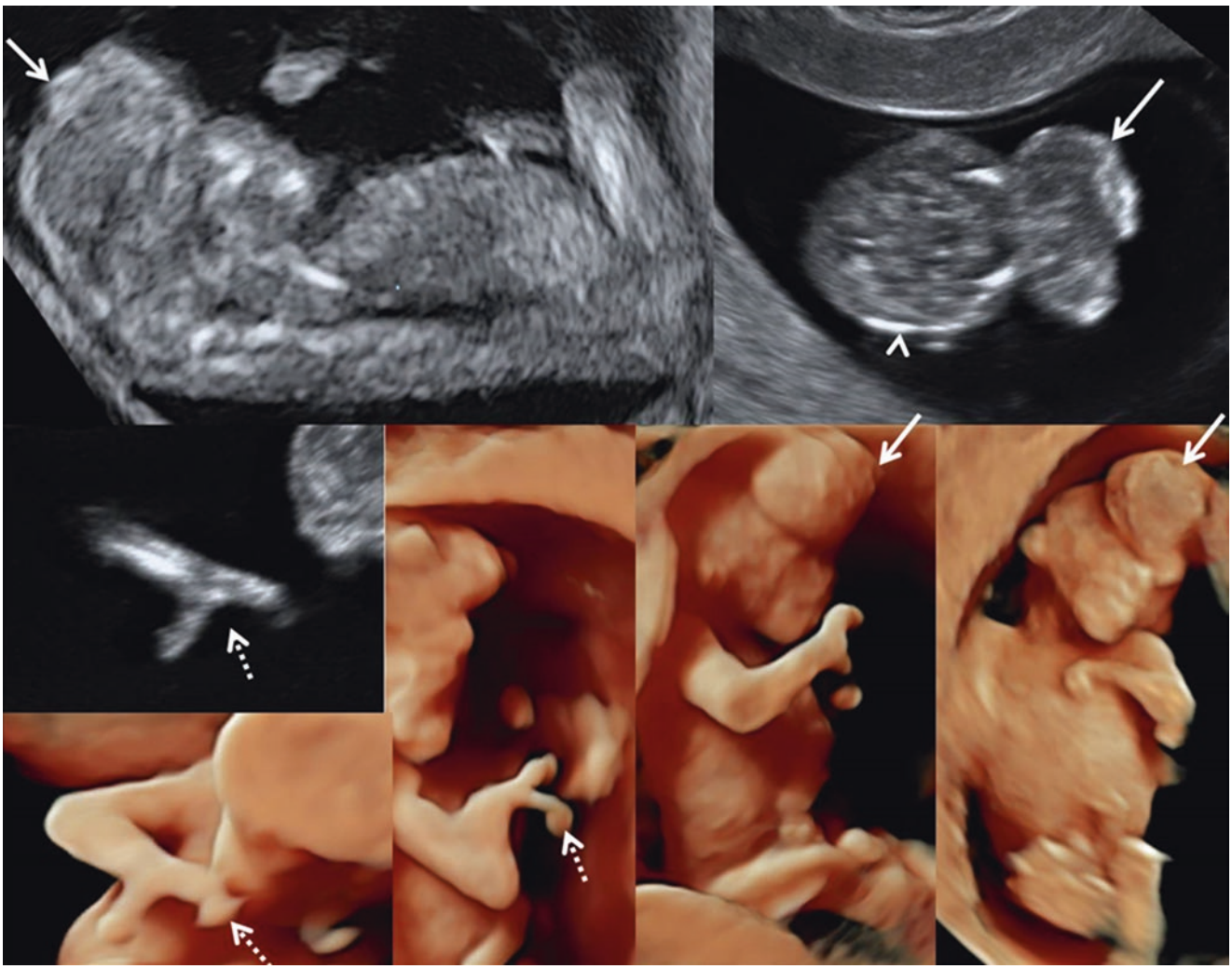

Fig. 8.13 12 weeks (TVS) *frontal encephalocele with bilateral elbow flexion contractures and right oligoectrodactyly* – midsagittal section of the fetus, axial section of the cranium, coronal section of the right hand and 3D surface rendered images of fetus – frontal encephalocele (solid arrows), right upper limb oligoectrodactyly (dotted arrows), calvarial vault bones (arrowhead)

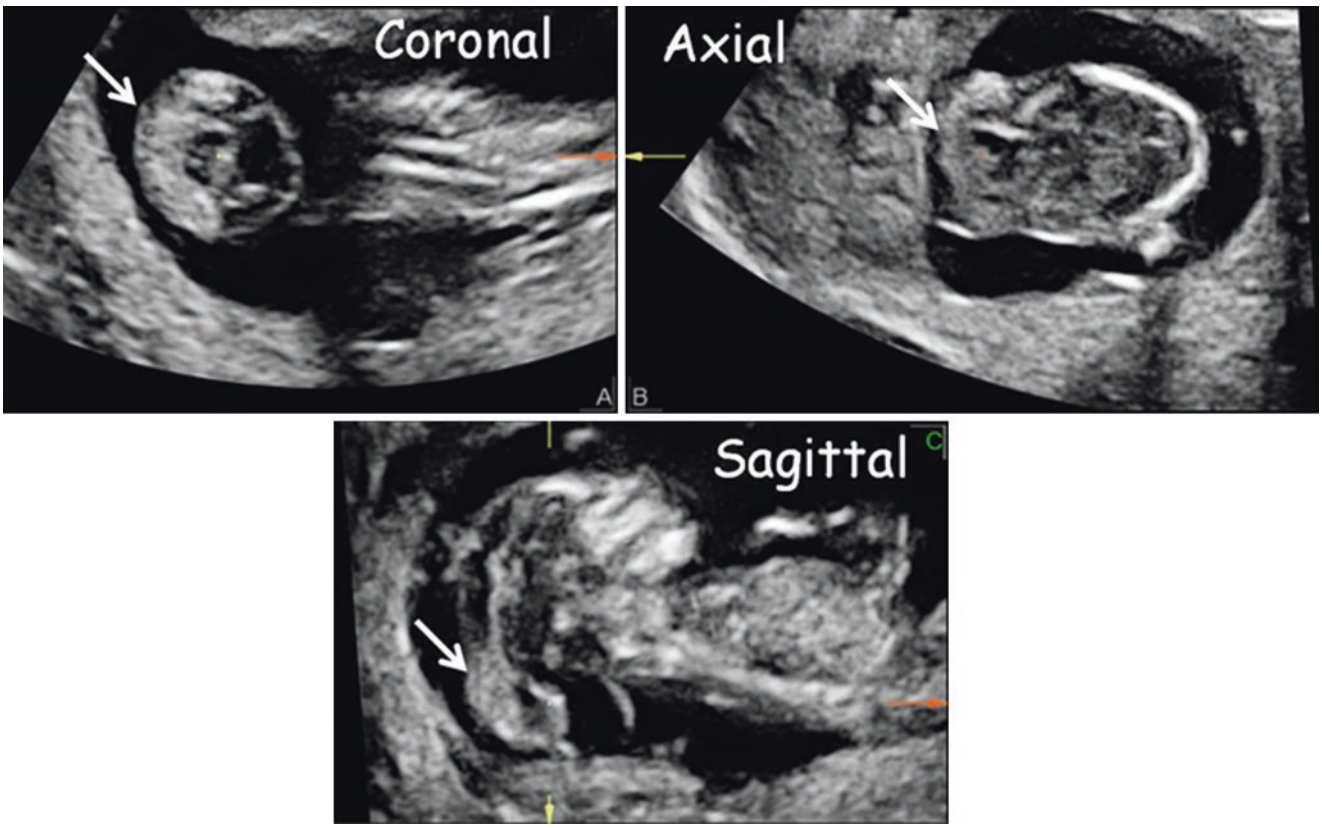

Fig. 8.14 12 weeks (TAS 3D multiplanar display) *occipital encephalocele* – thin-walled cyst arising from the occipital region with brain content (solid arrow)

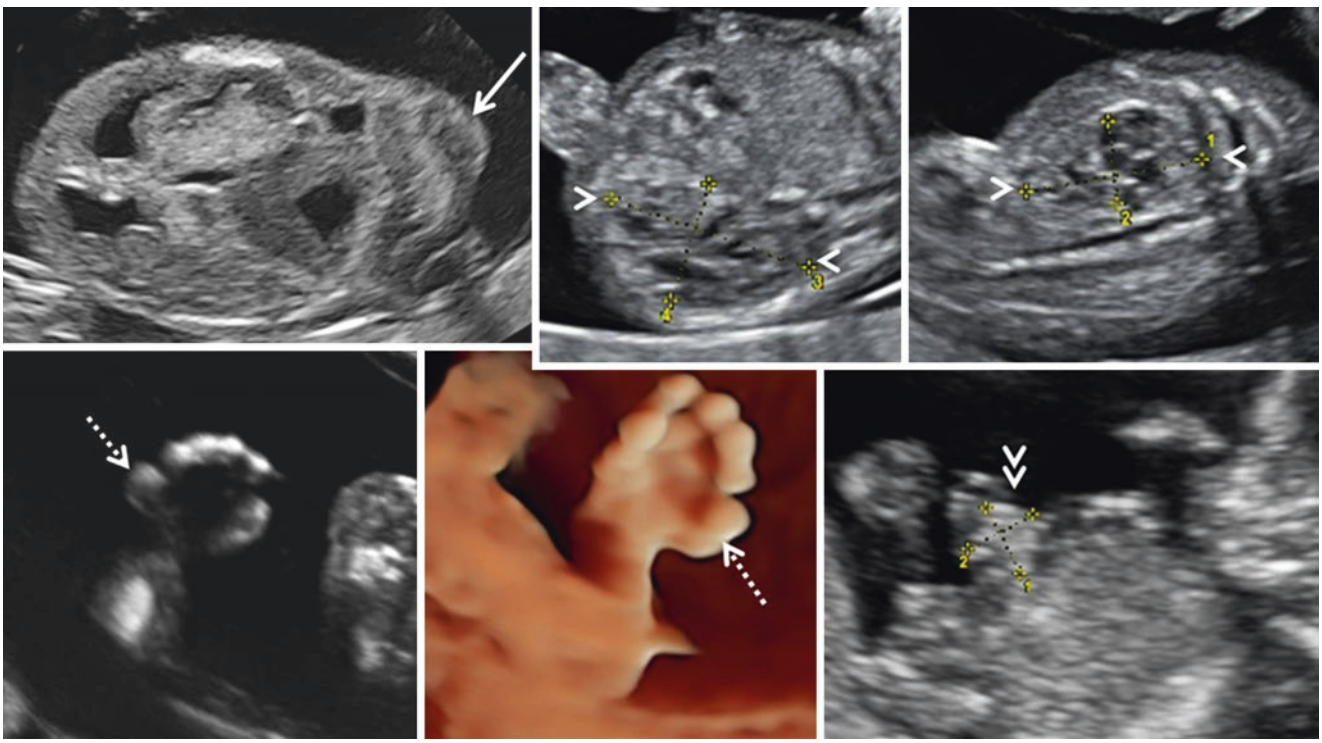

Fig. 8.15 12 weeks (TVS and 3D US) *Meckel-Gruber syndrome* – axial transventricular section of the cranium, axial and coronal sections of the abdomen, coronal section of the hand with 3D surface rendering, section through umbilical cord insertion to the abdomen – occipital encephalocele (solid arrow), bilateral cystic dysplastic kidneys (arrowheads), postaxial polydactyly (dotted arrows), omphalocele (double arrowheads)

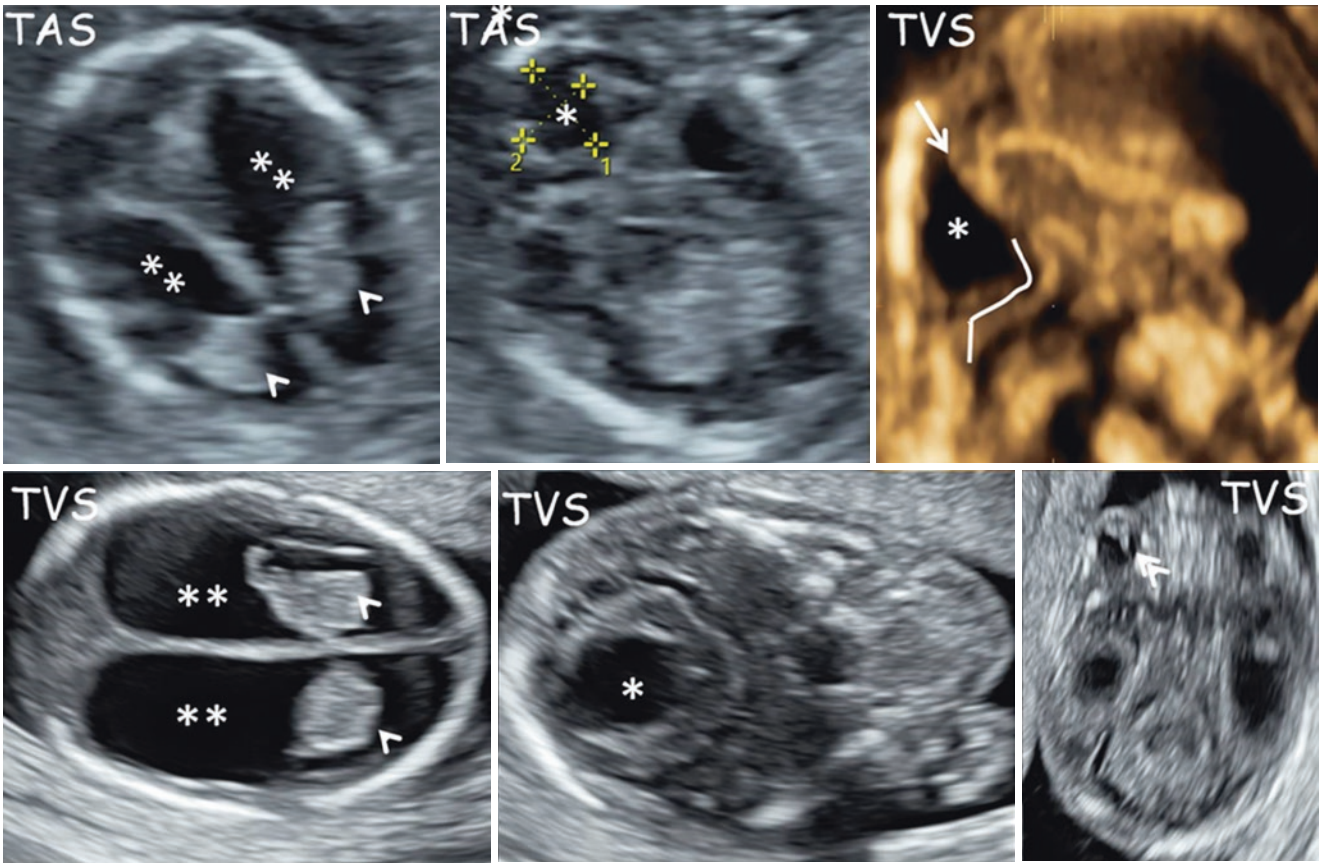

Fig. 8.16 13 weeks (TAS & TVS) *Walker-Warburg syndrome* – Axial transventricular and transcerebellar, 3D VCI midsagittal, axial transventricular and transcerebellar and frontal axial orbital sections - Bilateral severe lateral ventriculomegaly (**) with small, choroid plexuses separated from the lateral ventricular walls by CSF (arrowheads), posterior fossa cyst (*), vermis (arrow), direction of the arrow represents tentorial elevation (DWM), nonlinear line traces the Z shaped brainstem, non-attached retina (double arrowheads)

3. Size of the lesion varies from few millimetres to almost the size of the cranium.
4. Calvarial defect can be seen by transvaginal ultrasonography.
5. There is associated lateral ventriculomegaly especially if the brain is herniated.
6. In cases of abnormal IT with normal spine on transvaginal examination, an occipital cephalocele must be suspected and should be looked for (Figs. 8.5 and 8.6).
7. Associated abnormalities should prompt the diagnosis of a syndrome such as Meckel-Gruber (Fig. 8.15), Walker-Warburg (Fig. 8.16) and Joubert syndromes.

8.5 Holoprosencephaly

Failure of cleavage of the prosencephalon in the sagittal plane results in holoprosencephaly (HPE). The forebrain (cerebrum and basal ganglia) fails to divide into the right and left cerebral hemispheres and thalami. In alobar HPE there is complete failure to cleave. Partial cleavage results in semilobar and lobar HPE. Alobar HPE is lethal. It can be diagnosed in the 11–14 weeks examination.

The ultrasound findings are as follows:

1. The cranial midline (falx) echoes are not seen in the axial section. The 'butterfly' sign is absent (Figs. 8.17, 8.18 and 8.19).
2. Single midline primitive ventricle (monoventricle) extending from side to side is seen in the coronal section.
3. The thalamus is uncleaved.
4. Midline facial anomalies including hypotelorism, cyclops, proboscis, absent nose and median cleft lip are seen (Figs. 8.17, 8.18 and 8.19).
5. Premature or accelerated ossification of the frontal bones and narrowing of the metopic suture may be seen (Fig. 8.19).
6. Presence of extracraniofacial anomalies such as polydactyly, cardiac defects, omphalocele and hyperechoic kidneys may point towards a chromosomal abnormality (trisomy 13, trisomy 18, triploidy) or syndrome (Rubinstein-Taybi, Meckel-Gruber) (Figs. 8.17, 8.18 and 8.19).
7. HPE can be of autosomal dominant non-syndromic origin. The abnormal gene may not manifest in the parent with the mutation. Recurrence of HPE in subsequent pregnancies may occur.

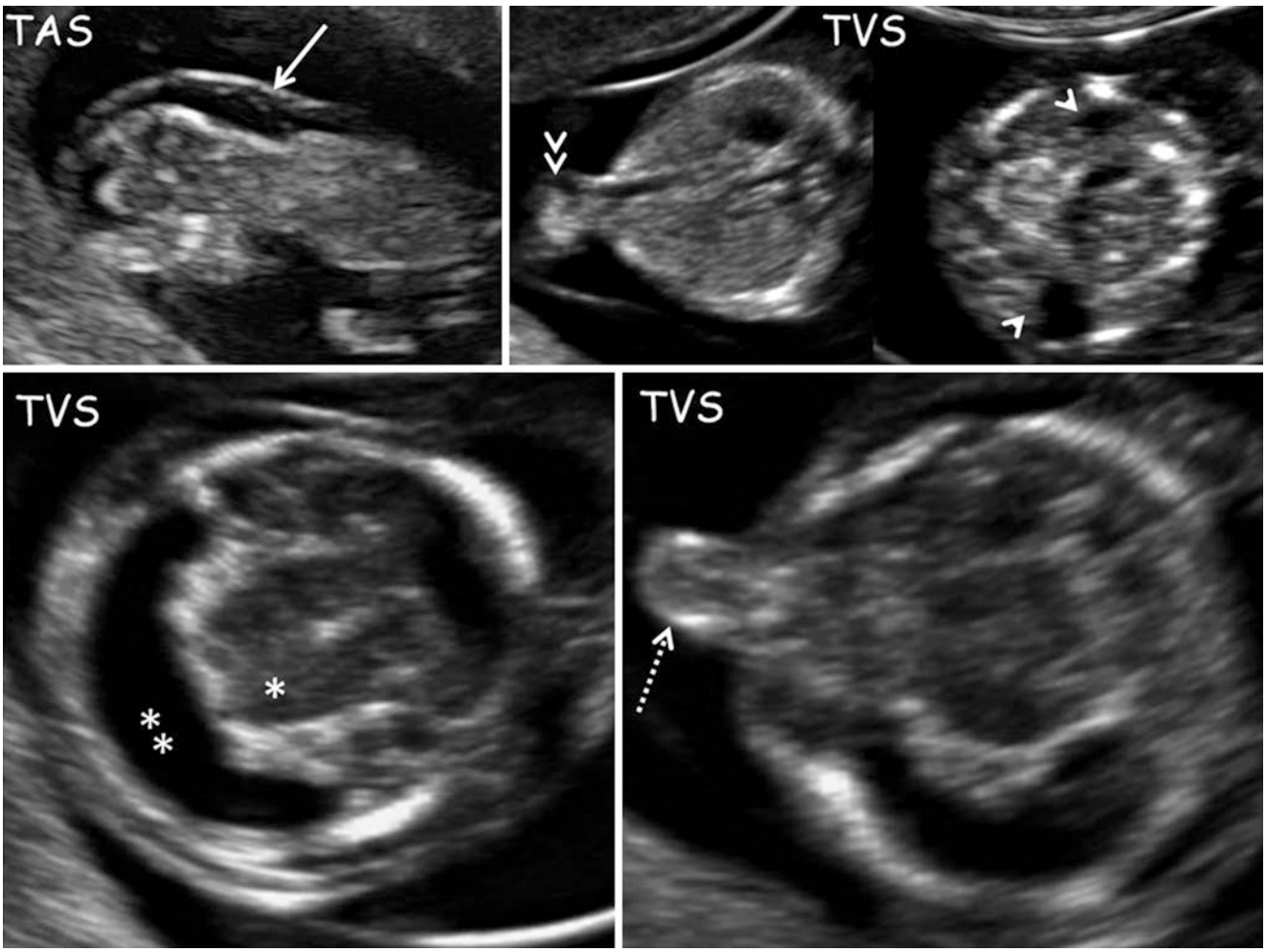

Fig. 8.17 12 weeks (TAS and TVS) *alobar holoprosencephaly with extracraniofacial anomalies* – sagittal section of fetus, axial section at cord insertion site into the abdomen, axial section of the neck, semi-coronal section of the cranium, axial section of the face at the level of the forehead – dorsal oedema (solid arrow), omphalocele (double arrowheads), bilateral dilated jugular sacs (arrowheads), uncleaved primitive monoventricle (**), uncleaved thalamus (*), proboscis (dotted arrows)

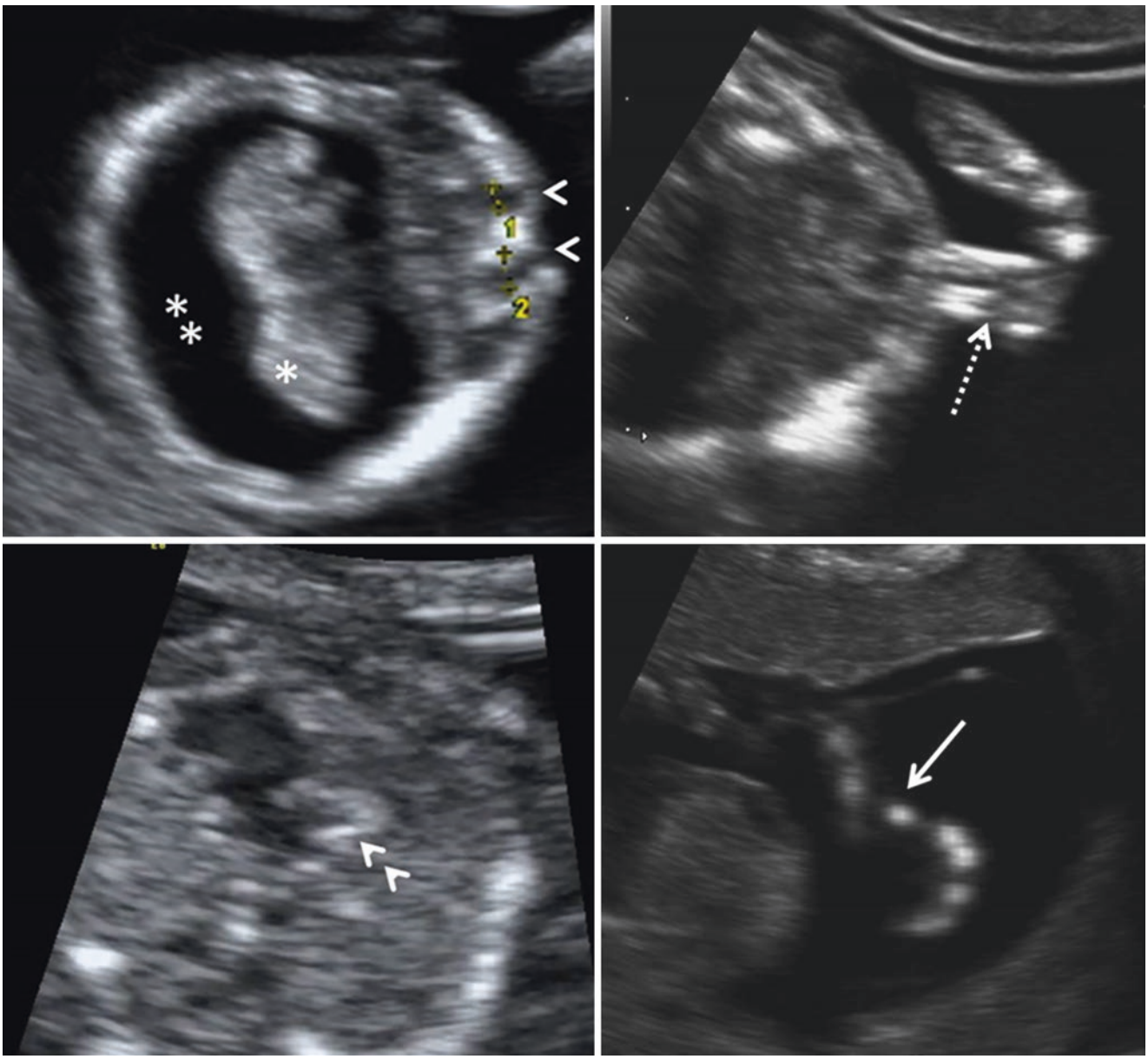

Fig. 8.18 12 weeks (TVS) *alobar holoprosencephaly with extracraniofacial anomalies* – semicoronal section of the cranium, axial section of the face at the level of the forehead, four chamber view of the heart, axial section of the hand – uncleaved primitive monoventricle (**), choroid plexus running across from side to side (*), bilateral microphthalmia and hypotelorism (arrowheads), proboscis (dotted arrows), hypoplastic left heart (double arrowheads), polydactyly (solid arrow)

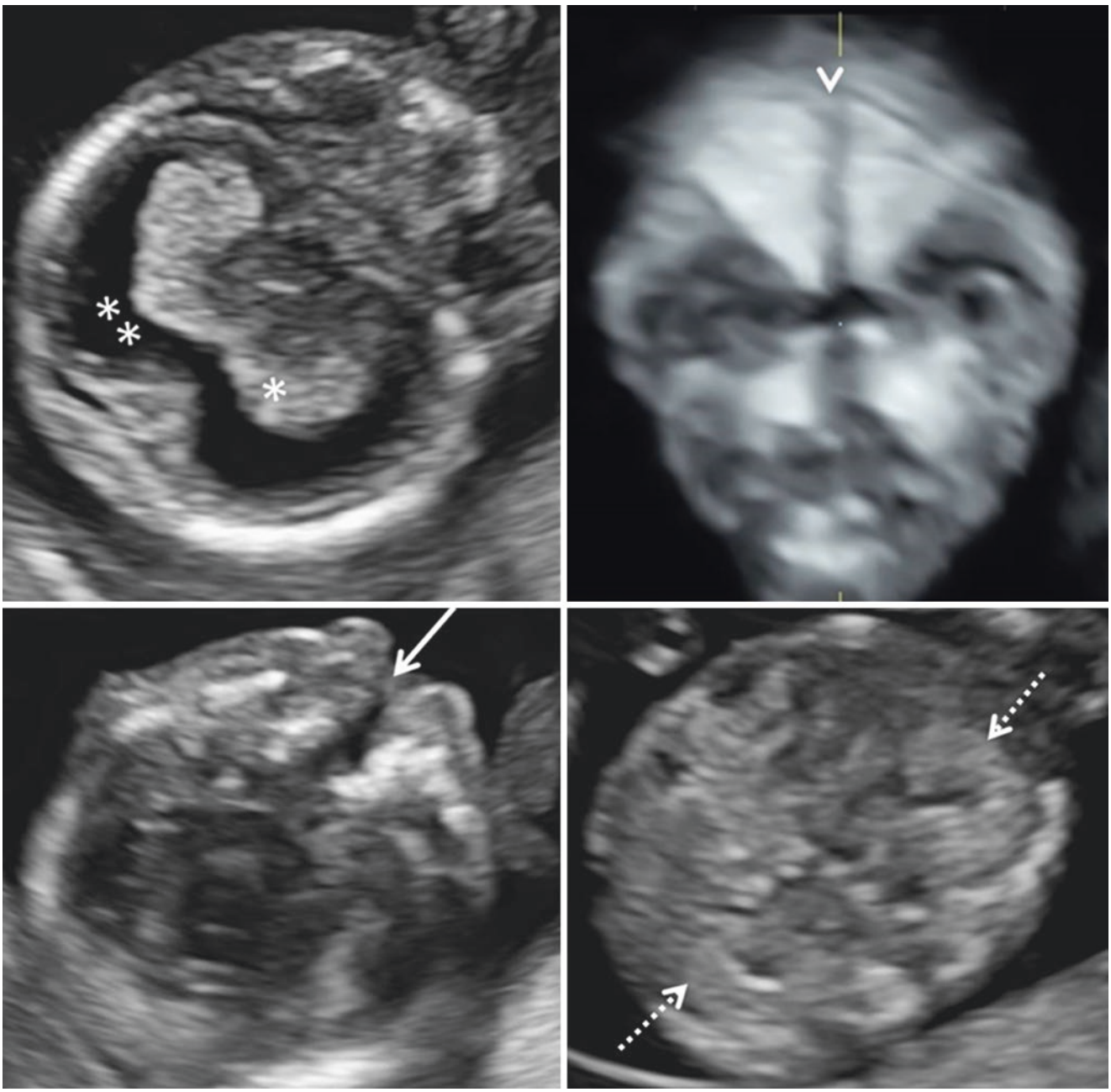

Fig. 8.19 13 weeks (TVS) *alobar holoprosencephaly with extracraniofacial anomalies* – semicoronal section of the cranium, 3D coronal face rendered image, axial section through the upper lip and jaw, axial section of the abdomen – uncleaved primitive monoventricle (**), choroid plexus running across from side to side (*), narrow metopic suture (arrowhead) due to premature or accelerated ossification of frontal bones, median cleft lip and palate (solid arrow), bilateral hyperechoic kidneys (dotted arrows)

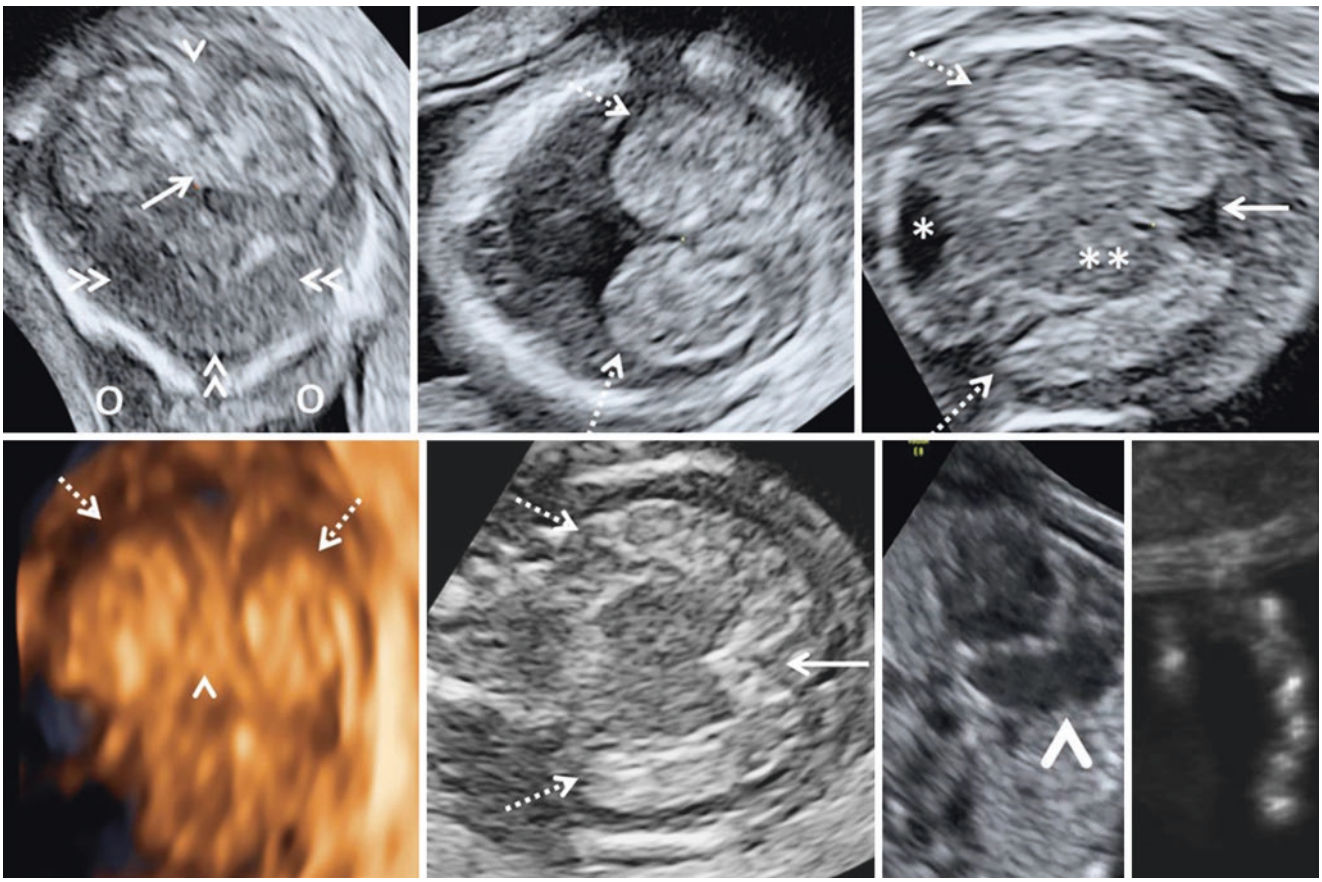

Fig. 8.20 12 weeks (TVS and 3D US) *lobar holoprosencephaly with extracraniofacial anomalies* – coronal transfrontal, axial transthalamic and transcerebellar, 3D rendered coronal transcerebellar and semicoronal sections of the cranium, four-chamber view and axial section of the left hand – uncleaved anterior horns of the lateral ventricles with choroid plexus continuous across the midline (solid arrow), inferior frontal cerebral cortex is continuous across the midline with no IHF (double arrowheads), cleaved body and posterior horns (dotted arrows) posterior fossa cyst (*), uncleaved thalamus (**), IHF (arrowhead) seen with cleaved occipital lobes, single inlet single ventricle (large arrowhead), postaxial polydactyly

Lobar and semilobar holoprosencephaly may be occasionally diagnosed in the 11–14 weeks US examination (Fig. 8.20).

8.6 Ventriculomegaly

The lateral ventricles do not increase beyond 10 mm in the first trimester. Increased fluid in the lateral ventricles manifests as follows:

1. The choroid plexus is thin, filling less than half the diameter of the lateral ventricle. The ratio of choroid plexus length and area to that of the lateral ventricle may aid in the diagnosis (Figs. 8.16 and 8.21).
2. Separation of the choroid plexus from the medial and lateral walls of the lateral ventricle.
3. Appearance of excessive fluid in the lateral ventricle is noted.
4. In aqueductal stenosis, a dilated third ventricle may be seen.

Invasive testing for fetal karyotyping / chromosomal microarray and repeat US examinations at 15 and 20 weeks are recommended.

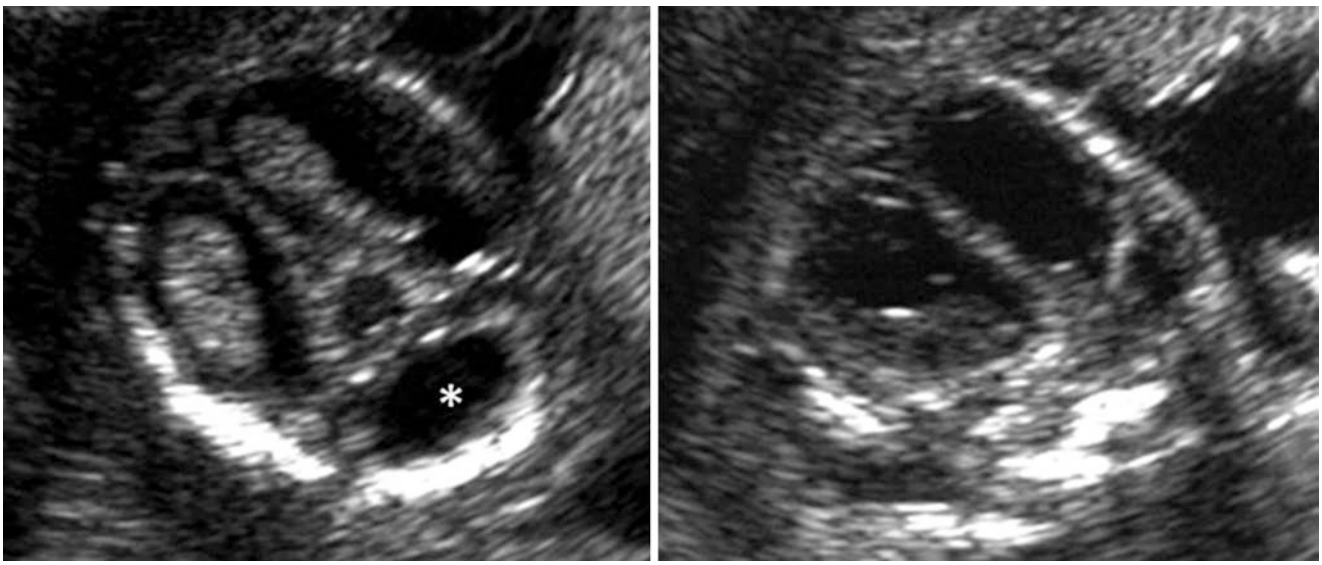

Fig. 8.21 12 weeks (TAS) *lateral ventriculomegaly with posterior cranial fossa cyst* – axial transcerebellar section and coronal transfrontal sections – choroid plexus appears discrete and separated from the lateral ventricular margins, the lateral ventricular fluid content appears to be excessive, posterior cranial fossa cyst (*)

Suggested Reading

1. Finn M, Sutton D, Atkinson S, Ransome K, Sujenthiran P, Ditcham V, Wakefield P, Meagher S. The aqueduct of Sylvius: a sonographic landmark for neural tube defects in the first trimester. Ultrasound Obstet Gynecol. 2011;38:640–5.
2. Lachmann R, Chaoui R, Moratalla J, Picciarelli G, Nicolaides KH. Posterior brain in fetuses with open spina bifida at 11 to 13 weeks. Prenat Diagn. 2011;31:103–6.
3. Chaoui R, Benoit B, Mitkowska-Wozniak H, Heling KS, Nicolaides KH. Assessment of intracranial translucency (IT) in the detection of spina bifida at the 11-13-week scan. Ultrasound Obstet Gynecol. 2009;34:249–52.
4. Volpe P, Contro E, Fanelli T, Muto B, Pilu G, Gentile M. Appearance of fetal posterior fossa at 11-14 weeks in fetuses with Dandy-Walker malformation or chromosomal anomalies. Ultrasound Obstet Gynecol. 2016;47:720–5.
5. Chaoui R, Benoit B, Mitkowska-Wozniak H, Heling KS, Nicolaides KH. Assessment of intracranial translucency (IT) in the detection of spina bifida at the 11-13-week scan. Ultrasound Obstet Gynecol. 2009;34:249–52.
6. Chaoui R, Nicolaides KH. Detecting open spina bifida at the 11-13-week scan by assessing intracranial translucency and the posterior brain region: mid-sagittal or axial plane? Ultrasound Obstet Gynecol. 2011;38:609–12.
7. Maruotti GM, Saccone G, D'Antonio F, Berghella V, Sarno L, Morlando M, Giudicepietro A, Martinelli P. Diagnostic accuracy of intracranial translucency in detecting spina bifida: a systematic review and meta-analysis. Prenat Diagn. 2016;36:991–6.