Correlation of abnormal findings on fetal MRI with clinical and pathologic findings

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Aims and objectives

To show the anomalies observed on fetal MRI together with clinical, surgical, and/or pathologic images of these anomalies. To evaluate the role of fetal MRI in prenatal diagnosis.
Methods and materials

Since 1997 we have studied 1025 pregnant women by MRI at our center; 277 fulfilled the following inclusion criteria: availability of fetal MRI as well as clinical, surgical, or pathologic images and/or reports that can be correlated with the findings on MRI.

All studies were performed because an anomaly had been detected with US and were performed during the same gestational week whenever possible.

The MRI studies were done using multiple-element body coils on 1 and 1.5T scanners. We acquired images using SS-HF-RARE or HASTE sequences; occasionally, we also obtained single volumetric images with SS-RARE sequences and T1-weighted fast gradient-echo images. No special preparations were necessary for the woman or the fetus and the entire MRI examination took about 20 minutes.
Results

In the 277 pregnant women with a total of 288 fetuses included in the study, there were 435 anomalies (Table 1 on page 19): 164 craniofacial and neural tube anomalies (77 central nervous system, 65 head and neck and 22 neural tube and spine anomalies), 57 thoracic anomalies, 83 abdominal anomalies (divided in 47 abdominal 36 and genitourinary anomalies), 98 anomalies of the limbs and soft tissues, and 33 other anomalies; MRI failed to detect 164 anomalies. Pregnancy was voluntarily interrupted in 124 fetuses and 13 fetuses were stillborn.

This presentation shows the most representative cases in each category as well as a brief discussion of each one in order to illustrate the correlations between the fetal MRI findings and the clinical and pathologic findings.

1. CENTRAL NERVOUS SYSTEM, HEAD AND NECK AND NEURAL TUBE AND SPINE:

The development of the central nervous system (CNS) is a complex and continuous process that begins in the early fetal life. The knowledge of this process is essential to understand the congenital anomalies of the SNC and spine, but a detailed discussion of the neuroembryology is beyond the scope of this study.

There are numerous congenital malformations of the CNS and craniofacial structures, and more than 2,000 reported anomalies. One third of all fetal anomalies affect the CNS and more than 75% of all dead fetuses have a CNS-related malformation. In recent years, MR imaging has been established as an excellent alternative to ultrasound (US) when US is limited or inconclusive. In our study, CNS and craniofacial anomalies represent 37% of all anomalies. We have divided them in:

1.1 Central Nervous System (CNS): Table 2 on page 19 and Figures 1-15.

1.1.1 Midline Anomalies:

- **Holoprosencephaly**: Genetically and phenotypically heterogeneous disorder that is usually severe and affects the development of the brain and face. It is a result of a failure of division of the midline structures, especially the lateral ventricles and cerebral hemispheres. There are three varieties of holoprosencephaly: alobar (Fig. 1 on page 20)(the most severe; there is a dorsal cyst and hydrocephalus that cause macrocephaly,
and the interhemispheric fissure and the falx cerebri are totally absent), semilobar (Fig. 2 on page 21, Fig. 3 on page 22) (the two cerebral hemispheres are partially separated posteriorly with absent septum pellucidum, caudate heads are fused in midline, and partial occipital and temporal horns) and lobar type (Fig. 4 on page 23) (brain is almost completely divided with absent septum pellucidum, formed lateral ventricles but dysplastic frontal horns). The most important findings on MR are a single primitive ventricle and fused thalami. Dilatation of this monoventricle may be obvious later, but is often mild in the early stages of pregnancy. Facial anomalies are very frequent (cyclopia, hypotelorism, anophthalmia, arhinia and central cleft lip).

- **Agenesis of the corpus callosum** (Fig. 5 on page 24, Fig. 6 on page 25): The incidence is 0.3-0.7% in the general population and its etiology is multifactorial (genetic and teratogens are the most significant). This anomaly can be subclinical or associated with mental disability. The defect of development of the corpus callosum (a broad plate of dense myelinated fibers that interconnects both hemispheres) can be partial or total, and it usually takes place between 12 and 18 weeks' gestation. The MR findings are: ventriculomegaly with disproportionate enlargement of the atria and occipital horns, prominent interhemispheric fissure and increased separations of the frontal horns. There is also alteration of the circumvolutions which show a radial disposition. Agenesis of the corpus callosum can be isolated or in combination with other CNS anomalies including frontal encephalocele, alobar holoprosencephaly, microcephaly, intracranial lipomas, interhemispheric cysts, neural tube defects, posterior fossa anomalies and others.

- **Septo-optic dysplasia** (Fig. 7 on page 26): Absence of the septum pellucidum and optic disc hypoplasia. This anomaly has been regarded as part of the spectrum of the holoprosencephalies because of its association with midline defects. The optic nerves and chiasm are affected by different degrees of hypoplasia and the pituitary infundibulum may be absent. MRI shows absence of the cavum septi pellucidi with central fusion of frontal horns. The corpus callosum may be normal but is frequently described as thinned in postnatal follow-up studies. Ventriculomegaly may be present also.

### 1.1.2 Ventricular System. Hydrocephalus:

Ventriculomegaly is frequently the consequence of a cranial or cerebral malformation or disruptive event (for example in cerebral hemiatrophy (Fig. 8 on page 27), but may be secondary to a number of etiologies, intra and extracranial, chromosomopathies and congenital infections. Ventriculomegaly is considered when the distance between the lateral ventricles at the level of the atrium is superior to 10 mm at 2\textsuperscript{nd} trimester and hydrocephalus when it is more than 15 mm. When the ventriculomegaly is not associated with other anomalies it usually has good prognosis. The prognosis of the hydrocephalus depends on its degree and etiology.
1.1.3 Abnormal neuronal migration: The disorders of migration are characterized by the incomplete formation of the cortical layers, with abnormal locations of neurons that have failed to reach their final destination. Macroscopically, the main finding is an alteration in the convolutional pattern of the brain, which may be associated with modifications in brain mass and size of the ventricles (ventriculomegaly). These anomalies include: lissencephaly (Fig. 9 on page 28) (absence of convolutions), polymicrogyria (increased number of convolutions), schizencephaly (Fig. 10 on page 29) (gray matter-lined cleft extending from the ventricle to the subarachnoid space) and heterotopias.

1.1.4 Posterior Fossa abnormalities:

- **Chiari II: Chiari malformations** represent a broad group of anomalies that affect the CNS midline and spinal cord. The common pathogenesis is an underdevelopment of the posterior fossa. Prenatal diagnosis of type I Chiari (caudal descent of the cerebellar tonsil) is exceedingly rare in the literature. However, Chiari type 2 cases (where there is displacement of the medulla, fourth ventricle and cerebellar vermis through the foramen magnum), can be diagnosed prenatally by US and MR. MR may be useful to characterize the affected structures, the degree of displacement the degree of downward displacement of the structures and the most common associated anomalies such as hydrocephalus and spinal meningocele (Fig. 26 on page 47, Fig. 27 on page 48, Fig. 28 on page 49).

- **Dandy-Walker malformation (Fig. 11 on page 30):** Related abnormalities are Dandy-Walker variant and mega cisterna magna. This syndrome includes ventriculomegaly and vermian agenesis with communication between the 4th ventricle and an enlarged cisterna magna. There are other similar posterior fossa abnormalities that have been included in the Dandy-Walker complex. Dandy-Walker malformation has an estimated prevalence of 1 per 30,000 births and is very associated with other fetal anomalies and chromosomopathies.

- **Cerebellar hypoplasia:** Cerebellar hypoplasia may be global or focal (affecting a specific cerebellar structure). The imaging findings of global cerebellar hypoplasia show a cerebellum with normal or near-normal shape, but with a reduction in the volume and prominence of the subarachnoid. Unilateral cerebellar hypoplasia is usually secondary to prenatally disturbances, and the main finding is an asymmetry in the size of the cerebellar hemispheres. Another posterior fossa disorder is the inferior vermian hypoplasia which may be isolated; fetal MRI provides a more sensitive and specific diagnosis of inferior vermian hypoplasia compared with US.

- **Rhombencephalosynapsis (Fig. 12 on page 31):** Total or partial absence of the cerebellar vermis and fusion of the cerebellar hemispheres. It
may be isolated or combined with other fetal abnormalities. The MRI findings are agenesis or hypogenesis of the vermis and continuity of the cerebellar hemispheres, which creates a horseshoe-shaped arch across the midline.

1.1.5 Tumors: intracranial tumors are rare and its incidence has been estimated at 0.34 per 1 million live births. Embryonic tumors and specially teratomas are by far the most frequent variety diagnosed antenatally. The final diagnosis is almost impossible at imaging. In our study there is a case of a fetus with a primitive neuroectodermal tumor (PNET) (Fig. 13 on page 32) (this term no longer appears in the current WHO classification of CNS tumors), an aggressive neoplasms that originates from the neural crest. It is associated with hydrocephalus, macrocephaly and polyhydramnios probably related to failure of swallowing, whether this is neurologically induced or is the consequence of mechanical obstruction to the pharynx. The most important differential diagnosis is with intraparenchymal hemorrhage.

1.1.6 Cysts: Interhemispheric cyst (Fig. 14 on page 33): Cystic lesions are one of the most frequent findings of the fetal brain. In our study we recorded a glioependymal cyst: they are cystic lesions with an ependymal lining, and they may be intra or extra-axial. Fetal MRI demonstrates T2 hyperintense lesions; that may be difficult to distinguish them from the more frequent arachnoid cysts; although glioependymal cyst are more frequently located at the interhemispheric region and is associated with agenesis of the corpus callosum.

1.1.7 Intracranial hemorrhage (Fig. 15 on page 34): Antenatal fetal intracranial hemorrhage usually originates in the subependymal germinal matrix region (as in the preterms) and it may occur spontaneously or occur in association with various maternal and fetal conditions. Predisposing fetal conditions at risk include tumors and co-twin demise and predisposing maternal conditions include coagulation disorders and drugs. MRI is very useful because is very sensitive to small hemorrhages and can help to distinguish them from brain tumors; the T2 signal intensity may vary depending on the chronicity of the hemorrhage.

1.1.8 Hydranencephaly (Fig. 16 on page 35): is the absense of the cerebral hemispheres which are replaced by a sac-like structure containing CSF. It is though to derive from a destructive process that leads to liquefaction of the cerebral hemispheres with subsequent cavitation and resorption of the necrotized tissue, filling the cavity with CSF. Fetal hypoxia and congenital infections such as toxoplasmosis and cytomegalovirus have been associated with hydranencephaly. It may be difficult to differentiate from extreme hydrocephalus and alobar holoprosencephaly.
1.2 Head and Neck: Table 3 on page 36 and Figures 16-21.

1.2.1 Cleft lip and cleft palate (Fig. 17 on page 37, Fig. 18 on page 38, Fig. 19 on page 39): Alteration of the fusion of the fronto-nasal and maxillary processes and the primary and secondary palatine processes. Cleft lip and cleft palate can occur separately or together. The overall incidence is 0.15 % of all births, which represents 7.5 % of all fetal anomalies. Predisposing factors include rubella, medication, alcohol, tobacco, and drug abuse. These anomalies are sometimes difficult to diagnose and this is especially true of cleft palate.

1.2.2 Tumors and masses:

- Epignathus (Fig. 20 on page 40): The incidence for epignathus is approximately 1 per 35,000 live births. Associated anomalies are seen in about 6% of epignathus and include facial clefts, bronchial cysts, hypertelorism and cardiopathies.
- Granulosa cell tumor (epulis)(Fig. 21 on page 41): It is a rare, benign pedunculated tumor arising anteriorly from the maxillary alveolar ridge. There are published reports of epulis in a triple X female fetus.
- Hemangioma and cystic lymphangioma: They are the most common tumors of infancy and the majority of them is found at birth. Lymphangiomas are common in the neck, and they can spread to the chest wall.
- Goiter (Fig. 22 on page 42): Enlargement of the thyroid can result from either hypo- or hyperthyroidism. The global incidence of congenital hypothyroidism is of 1 per 3,700 live births. The incidence of fetal thyroid dysfunction in maternal Graves disease is 2-12%.

1.3 Neural tube and spine: Table 4 on page 43 and Figures 22-19. Neural tube defects (NTD) are a heterogeneous group of anomalies that result from failure of normal neural tube closure between the third and fourth week of embryologic development. The most frequent forms are anencephaly, encephalocele and spina bifida; less common types are iniencephaly (Fig. 33 on page 55) and amniotic band syndrome.

1.3.1 Encephalocele (Fig. 23 on page 44): Cephaloceles or encephaloceles are protrusions of intracranial structures through an osseous defect in the skull. Most commonly these defects are located at the midline of occipital and frontal regions, but they can also be parietal, nasal and sphenoidal. MR is useful for detecting small lesions and the type of herniation. Encephaloceles are often part of specific syndromes, and also very associated with ventriculomegaly and spina bifida.

1.3.2 Spina bifida: Congenital defect of the vertebrae resulting in exposure of the contents of the neural canal. In the vast majority of cases the defect is localized to the dorsal arch of the vertebrae, and rarely of the vertebral body. It is most commonly found
in the lumbar region, but it can occur anywhere in the spine. The lesions are commonly divided into ventral and dorsal defects; dorsal defects are by far the most common, and they are subdivided into two types:

- **Closed (occulta):** represents 15% of the cases and is characterized by a small defect completely covered by skin (*Fig. 24 on page 45*).
- **Open (aperta):** is the most frequent lesion (85%). The neural canal may be exposed or a thin membrane may cover the defect. More often the lesion looks like a cystic tumor (spina bifida cystica). If the tumor contains only meninges, the lesion is referred to as **meningocele**; more commonly the lesion contains also neural tissue, known as **myelomeningocele**. The term myeloschisis is used when the neural canal is widely opened and is part of the wall of the tumor. There is always a defect in the vertebrae. Associated anomalies are common, specially CNS defects and lower extremities malformations (*Fig. 25 on page 46 Fig. 26 on page 47 Fig. 27 on page 48 Fig. 28 on page 49*).

### 1.3.3 Sacrococcygeal teratoma (*Fig. 29 on page 50*)

*germ cell tumor arising from the presacral area. There are four types of these tumors:

- **Type 1:** External with minimal presacral component.
- **Type 2:** Predominantly external with significant intrapelvic component.
- **Type 3:** Predominantly internal with abdominal extension.
- **Type 4:** Entirely internal with no external component.

They are seen as cystic, solid or mixed cystic and solid masses arising from the sacrococcygeal region. Depending on the type of teratoma, the MR can detect a cystic, solid or mixed cystic and solid mass in the sacrococcygeal region with or without pelvic component.

### 2. THORAX: Table 5 on page 51 and Figures 30-53.

Fetal survival after birth is extremely dependent upon adequate development of the lungs during fetal life. Pulmonary underdevelopment may be caused by decreased thoracic size, space-occupying lesions inside the chest or insufficient amniotic fluid volumes.

Heart defects comprise a group of malformations present in approximately 8 in 1,000 live births and 25% of them are associated with other malformations. Other thoracic anomalies (tracheo-thoracic, thoracic wall defects, tumors...) play an important role in childhood morbidity and mortality.
2.1 Reduction of the thoracic size: Pulmonary hypoplasia can be caused by a reduction in the size of the thoracic cavity. This is especially common in some types of skeletal dysplasia, such as thanatoporic dwarfism, Jeune's asphyxiating dystrophy, achondrogenesis, or type II osteogenesis imperfecta. These conditions are considered lethal due to pulmonary hypoplasia induced by the hypoplastic rib cage. Fetuses suffering from these conditions sometimes show polyhydramnios, possibly related to esophageal compression by the smaller-than-usual chest, and may suffer associated gastrointestinal anomalies or hypotonia.

- **Thanatophoric dysplasia** (Fig. 30 on page 52): The prevalence is 0.2-0.6 in 10,000 live births. Key findings are: severe micromelia, macrocrania, and severe pulmonary hypoplasia. Other associated anomalies include: holoprosencephaly, agenesis of the corpus callosum and ventriculomegaly, horseshoe kidneys, and heart defects.
- **Achondrogenesis**: The prevalence is 0.09-0.23 in 10,000 live births. It is characterized by micromelia, macrocrania, pulmonary hypoplasia, and reduced skeletal mineralization. As reduced skeletal mineralization cannot be seen on MRI, US plays an important role in distinguishing this anomaly from thanatophoric dysplasia.
- **Imperfect osteogenesis** (Fig. 31 on page 53): In this condition, collagen alterations cause fragile bones leading to multiple fractures. The estimated incidence is 1 in 60,000 live births. Six types have been described; type II is the lethal form affecting neonates. Thoracic hypoplasia with pulmonary hypoplasia is caused by shortened ribs and vertebral deformation due to fracturing.
- Other anomalies such as congenital hypophosphatasia (alkaline phosphatase deficiency) arthrogryposis (heterogeneous condition with contractures in multiple joints), chondrodysplasia punctate (autosomal recessive alteration caused by a peroxisomal alteration), amniotic band syndrome (Fig. 32 on page 54) (multiple bands that extend from the chorionic surface of the amnion to fetal tissues), iniencephaly (Fig. 33 on page 55) (a neural tube defect) or severe dorsal scoliosis (Fig. 34 on page 56) can be associated to thoracic hypoplasia and consequently to pulmonary hypoplasia.

2.2 Space-occupying lesions: They cause mass effect that leads to secondary pulmonary hypoplasia. The degree of pulmonary hypoplasia depends on the time this lesion appears, its duration and its severity. The most frequent are:

- **Diaphragmatic hernia**: The estimated incidence is 1-4.5 cases in 10,000 live births. Most cases (75-90 %) involve the left side (Fig. 35 on page 57) and the lesion is bilateral in 5% of all cases. Diaphragmatic hernia derives from failure of the pleuropitoneal canals to close, causing a defect through which abdominal contents can herniate into thoracic cavity. However, when the heart is displaced to the left (right-sided hernias, Fig. 36 on page 58) or when the stomach does not protrude through the
hernia (Fig. 37 on page 59), US diagnosis can be extremely difficult. In MRI, the distinct differences in signal intensity of the different tissues enables excellent discrimination among thoracoabdominal organs, even early in gestation, and makes it easy to diagnose diaphragmatic hernia. The prognosis is worse when the liver or its left lobe is herniated (Fig. 36 on page 58, Fig. 38 on page 60) because it results in mechanical problems and comprised fetal circulation through the ductus venosus. Some of them are associated to other fetal anomalies (Fig. 39 on page 61).

- **Congenital pulmonary airway malformations (CPAM):** Multicystic intrapulmonary mass with primitive lung tissue and bronchial and bronchiolarlike structures. Its incidence may be greater than previously believed because many cases are asymptomatic at birth (Fig. 40 on page 62). The most important prognostic factor is the size of the mass itself rather than the size of the cysts within it, as the larger the mass, the greater the resultant pulmonary hypoplasia (Fig. 41 on page 63). Pleural effusion is rarely associated except in cases of hydrops fetalis, which have a worse prognosis. CPAM is not usually associated to extrapulmonary anomalies. Sometimes the cysts have a connection to the tracheobronchial tree therefore they can increase their size progressively (Fig. 42 on page 64). It is not uncommon for the lesion to decrease in size during the third trimester and after birth. CPAM can be invisible on plain-film radiographs after birth and should therefore be studied by CT (Fig. 40 on page 62).

- **Pulmonary sequestration:** Pulmonary sequestration is a supernumerary lobe of the lung, separated from the normal tracheobronchial tree. Most lesions are located at the base of the left lung (Fig. 43 on page 65). Lesion size varies considerably, although small or moderate lesions tend to be more common. On MRI, the lesion is hyperintense on T2-weighted sequences (Fig. 43 on page 65, Fig. 44 on page 66, Fig. 45 on page 67). Sequestration is rarely associated with hydrothorax or hydrops fetalis. These lesions can decrease in size or even disappear during gestation or after birth. They can be associated to other anomalies such as diaphragmatic hernia or intestinal duplication (Fig. 45 on page 67). The prognosis is usually favorable. Approximately 10% of all extralobar sequestrations are located below the diaphragm; in these cases it is important to differentiate sequestration from neuroblastoma.

- **Bronchial atresia:** is an uncommon anomaly in which a segment of the bronchus does not communicate with the central airway. The portion distal to the atresia sometimes shows lesions similar to CPAM. It mostly affects the upper lobes (Fig. 46 on page 68) but it can affect more than one lobe lung (Fig. 47 on page 69) or both lungs. Peripheral bronchi are not depicted on MRI, but this entity can be suspected in cases with hyperintense areas in the lungs. It is difficult to differentiate from other pulmonary anomalies.

- **Bronchogenic cyst and others:** The bronchogenic cyst is a cystic lesion lined with bronchial epithelium. It can be found in the lung, mediastinum, and even in the neck or abdomen. **Congenital lobar emphysema** is caused by
anomalous development of a lobar or segmental bronchus or by bronchial compression causing bronchial dysplasia. MRI shows a hyperintense area of the lung and cannot differentiate this lesion from other pulmonary anomalies. **Neuroenteric cysts** consist of remains of enteric tissue that behave like cystic masses. They are normally located in the mediastinum and are commonly associated with vertebral anomalies.

- **Hydrothorax:** The accumulation of liquid in the pleural space is always pathologic. It is essential to determine whether the hydrothorax is isolated (most commonly, primary chylothorax) or forms part of hydrops fetalis (**Fig. 48 on page 70**). Unilateral affection usually represents chylothorax, and bilateral affection normally occurs in cases of hydrops fetalis.

**2.3 Inadequate volume of amniotic fluid:** A normal amount of amniotic fluid is essential to ensure proper development and maturation of the fetal lungs. Although different factors influence the regulation of amniotic fluid (fetal respiratory movements, fetal deglutition, uteroplacental blood flow, and others), fetal urine, especially after the 16th week of gestation, becomes the principal source of amniotic fluid. In general, oligohydramnios indicates poor renal function or a severe obstructive anomaly of the urinary system and subsequent fetal pulmonary hypoplasia that worsens the prognosis. Bilateral renal agenesis, autosomal recessive polycystic kidney disease (**Fig. 49 on page 71**), bilateral renal dysplasia, and severe obstruction of the lower urinary tract (**Fig. 50 on page 72**) are the genitourinary anomalies in which urine production is most severely affected, causing oligohydramnios and severe pulmonary hypoplasia.

**2.4 Cardiopathies:** Heart defects comprise a group of malformations present in approximately 8 in 1,000 live births. Although MRI is not well-suited to fetal heart studies, it does provide information regarding the size (**Fig. 51 on page 73**), shape and position of the heart. MRI also gives less accurate information about the great vessels of the mediastinum and heart tumors. The use of MRI is justified not only because of the transcendence of heart malformations themselves, but also because of the high incidence of associated anomalies, affecting up to 25% of cases (**Fig. 52 on page 74, Fig. 53 on page 75**).

**3. ABDOMEN:** **Table 6 on page 76** and Figures 54-69.

Congenital abdominal anomalies can occur in many fetal organs (the kidneys, ureters, bladder, genitals, gastrointestinal tract, liver, spleen, mesentery, and/or peritoneal cavity). The accurate determination of the location and morphological characteristics of an anomaly is essential for diagnosis. MRI’s ability to discriminate among tissues makes it a great help in the diagnosis of these pathologies. As some of these anomalies can be difficult to detect clinically at birth, prenatal diagnosis is important to ensure early intervention and minimize complications.
3.1 Gastrointestinal tract: We have divided them in superior and inferior tract.

3.1.1 Proximal:

- **Esophageal atresia**: congenitally interrupted esophagus is the most frequent disorder of this structure; it can be associated with tracheoesophageal fistula. MRI has been shown to increase diagnostic accuracy in establishing the diagnosis. The findings are polyhydramnios and small or empty stomach, and sometimes a large esophageal pouch in the fetal thorax is seen.

- **Duodenal obstruction**: Obstruction of the duodenum can be caused by atresia, stenosis, intraluminary diaphragm (Fig. 54 on page 77), annular pancreas (Fig. 55 on page 78), intestinal malrotation with Ladd's band (Fig. 56 on page 79), or midgut volvulus. The estimated incidence of these anomalies is 1/5,000 pregnancies. Approximately 50% of duodenal anomalies are associated to other anomalies (gastrointestinal, skeletal, cardiovascular, genitourinary, and chromosomal).

3.1.2 Small intestine and colon: Atresia of the small intestine affects approximately 1 in every 3,000-5,000 live births. Large bowel atresia is uncommon (less than 10% of all intestinal atresias). Vascular impairment during development is the most likely cause of this anomaly. Atresia most often affects the distal ileum or the proximal jejunum (Fig. 57 on page 80), and is multiple in approximately 6% of cases (Fig. 58 on page 81). While associated extraintestinal anomalies are rare, other intestinal anomalies (other atresias, malrotation, volvulus, duplication, gastroschisis, and meconium ileus) (Fig. 59 on page 82) are not. Anal atresia is more common; its incidence is estimated at 1 in 5,000 live births. In up to 70% of cases other anomalies (especially of the genitourinary tract) are associated (Fig. 54 on page 77).

3.2 Abdominal masses:

- **Enteric duplication cyst**: Duplications of the digestive tube are uncommon congenital abnormalities found anywhere along the alimentary tract from the tongue to the anus. Intestinal duplication is located on the mesenteric side of the gut and does not usually communicate with intestinal lumen. The most common location is the distal portion of the ileum, followed by the distal portion of the esophagus and the stomach. Although these lesions rarely cause intestinal occlusion in utero, they can cause intestinal occlusion and/or abdominal pain after birth, due to volvulus or invagination (Fig. 60 on page 83). Sometimes other anomalies such as bronchopulmonary sequestration are associated (Fig. 45 on page 67).
• **Mesenteric cyst:** They are considered to be lymphatic anomalies. They are often single, unilocular lesions, although they can be multilocular. They are sometimes mobile and change position.

• **Ovarian cyst:** This is a common cause of abdominal cysts in females during the prenatal period; it occurs in approximately 1 in 2,500 pregnancies. It usually presents during the third trimester (Fig. 61 on page 84). Excessive stimulation of the fetal ovaries by placental and maternal hormones seems to play a role in their etiology. Most are unilateral, although they can be bilateral. In fetuses they can be simple cysts or they can complicate with torsion and bleeding. They sometimes disappear in utero, though the majority disappear after birth.

• **Hepatic and splenic masses:** These lesions are very rare in the prenatal period. Cysts and hepatic epithelioid hemangioendotheliomas are the most common. They may appear within liver parenchyma or "hang" from its lower edge (Fig. 62 on page 85). They can be single or multiple. Masses in the spleen are almost always epidermoid cysts. They should be differentiated from duplication cysts, mesenteric cysts, urachal cysts, choledochal cysts, and ovarian cysts. Single cysts are not usually associated to other anomalies; however, we examined a fetus with a hepatic cyst and severe renal and cardiac anomalies (Fig. 63 on page 86).

### 3.3 Ascites:
Fetal ascites can be an isolated finding or it can be associated to hydrops fetalis (Fig. 64 on page 87). Isolated ascites is normally secondary to an intraabdominal problem rather than a systemic anomaly and it is often associated to fetal malformations. Genitourinary problems, usually from obstructive uropathy, are the most common cause. Other causes of isolated ascites are gastrointestinal in origin, usually meconium peritonitis, hepatic anomalies, heart defects, infections, metabolic disorders, and it is sometimes idiopathic.

### 3.4 Ventral Wall Malformations:

• **Gastroschisis** (Fig. 65 on page 88, Fig. 66 on page 89): Herniation of the abdominal viscera of the fetus into the amniotic cavity, secondary to a small defect affecting all of the layers of the abdominal wall. Associated anomalies are infrequent. Intestinal atresia or stenosis can be found in 7-30% of cases.

• **Omphalocele:** There is a midline defect of the abdominal muscles, fascia and skin that results in herniation of intraabdominal structures (Fig. 67 on page 90, Fig. 68 on page 91); the hernia is wrapped in a membrane of peritoneum. Other associated anomalies are present in approximately 54% of cases, the most common being heart defects, followed by CNS anomalies. Neural tube defects and genitourinary and gastrointestinal malformations. It is associated to trisomies 18 and 13. Beckwith-Wiedeman syndrome is often associated to omphalocele.
• **Inguinal hernia:** Is a rare cause of a scrotal mass. It is often an isolated anomaly and is more common in males than females. It results from a defect in the closure of the vaginal process (Fig. 69 on page 92).

4. **GENITOURINARY:** Table 7 on page 93 and Figures 70-77.

Fetal genitourinary disorders account for approximately 30% of all congenital abnormalities and are the most common intrauterine abnormalities detected prenatally. It is estimated that genitourinary anomalies are present in 1-3% of live births and account for 18% of all lethal congenital anomalies. Moreover, genitourinary malformations are a common cause of renal insufficiency in early life. Evaluation of the volume of the amniotic fluid provides important information about fetal renal function and uteroplacental flow. US is the best method for evaluating this, although MRI can acquire unique volumetric images using SS-RARE sequences that can evaluate the amount of amniotic fluid (subjectively and objectively). In our work we classify them in anomalies associated to good and poor prognosis:

4.1 **Anomalies associated with good prognosis:** Stable anomalies that do not compromise fetal viability, and have good prognosis after birth. The most common are: low-grade hydronephrosis, unilateral renal agenesis, horseshoe kidney and unilateral renal dysplasia (Fig. 70 on page 94). External genitalia anomalies (micropenis, hypospadias or ambiguous genitalia) can also be detected (Fig. 71 on page 95, Fig. 72 on page 96).

4.2 **Anomalies associated with poor prognosis:** Urinary tract anomalies associated with poor prognosis are those with oligohydramnios and/or pulmonary hypoplasia and those without oligohydramnios but associated with other severe malformations.

- **Bilateral renal agenesis:** is the complete absence of both kidneys, frequently associated with agenesis of the bladder, and is invariably lethal because of oligohydramnios and pulmonary hypoplasia. It has an approximate incidence of one in 4000 births and affects male fetuses more frequently. It is associated with craniofacial and limb anomalies. MR features of this entity include absence of the kidneys and bladder in addition to severe oligohydramnios.

- **Bilateral renal dysplasia and cystic diseases** (Fig. 73 on page 97, Fig. 74 on page 98): renal dysplasia, cystic diseases and urinary tract obstruction are the most common intrauterine malformations detected. They can be divided into three groups: polycystic kidney disease (recessive and dominant), dysplastic kidneys and syndrome associations of cystic kidney diseases; each one has its own characteristics. MR imaging shows variable-size kidneys with cystic lesions that can occupy the entire abdomen. Meckel-Gruber syndrome is a lethal autosomal recessive inherited syndrome with a 25% risk of recurrence. Polycystic kidneys, polydactylysm, and
encephalocele are the characteristic findings, and it is frequently associated with hepatic disease. Renal affection is similar to ARPKD (autosomal recessive polycystic kidney disease). Renal aplasia (aplastic displasia) represents the extreme form of renal dysplasia, with small and barely recognizable rudimentary kidneys (Fig. 75 on page 99).

- **Severe urinary tract obstruction (Fig. 76 on page 100, Fig. 77 on page 101):** They are easily detected because the urine is seen hyperintense on T2-weighted images. Pelviureteric junction obstruction is the most common cause of fetal upper urinary tract dilatation, but the prognosis is usually good. Anomalies with poor prognosis are posterior urethral valves and urethra agenesis.

### 5. LIMBS AND SOFT TISSUES: Table 8 on page 102 and Figures 78-97.

Unfortunately, few publications have focused on MRI of the fetal musculoskeletal system, therefore it has remained one of the least developed fields of fetal MRI. Congenital musculoskeletal disorders and malformations may occur in isolation or may be associated with other anomalies and syndromes. This work will only discuss the anomalies found on MRI that have been confirmed after birth. We have divided them in focal and diffuse anomalies.

#### 5.1 Focal Anomalies: They may range from an evident alteration such as absence of any limb (amelia), to supernumerary fingers (polydactyly).

- **Reduction Anomalies:** it can be characterized as involving the entire limb (micromelia) or a segment of it (Fig. 78 on page 103, Fig. 79 on page 104, Fig. 80 on page 105). Generally 50% of cases are isolated. Limb amputations may be caused by amniotic band syndrome (Fig. 81 on page 106), teratogens or vascular injury. Ectrodactyly (Fig. 82 on page 107) is the absence of one or more central fingers; it may be isolated or associated with facial defects and ectodermal dysplasia.
- **Polydactyly:** presence of more than five digits; they are classified as postaxial (more common) if the extra digits are on the ulnar or fibular side and preaxial if they are located on the radial or tibial side.
- **Positional anomalies: Clubfeet** or talipes equinovarus is by far the most common positional anomaly of the limbs; it characterized by a foot fixed in adduction, supination, and varus position (Fig. 83 on page 108, Fig. 84 on page 109, Fig. 85 on page 110). The prevalence among Caucasian population is 1-3 per 1,000 live births. The majority of cases (80%) they are isolated lesions; the most common associations are genetic disorders, chromosomopathies and CNS and spine abnormalities. This deformity may be caused by genetic (intrinsic) factors of the fetus or by environmental (extrinsic) factors such as reduced intrauterine space due to oligohydramnios or intrauterine lesions. Other less frequent positional
anomalies are **vertical astragalus** and **radial clubhand** due to radial hemimelia. *(Fig. 86 on page 111, Fig. 87 on page 112)*

- **Genu recurvatum** *(Fig. 88 on page 113)*: lower limb malformation with extension beyond neutral (more than 180º). It is usually bilateral.
- **Popliteal pterygium syndrome** *(Fig. 89 on page 114)*: a condition that affects the development of the face, skin, and genitals. There are skin folds in the knees, syndactyly, and excessive skin over the toenails.

### 5.2 Diffuse anomalies:

- **Skeletal dysplasias**: they are a large heterogeneous group of disorders that affect the development of multiple bones, causing abnormalities in their density, shape and size. Fetal skeletal dysplasias are difficult to diagnose in utero especially if there is no family history. Some of the factors that lead to difficulty in diagnosis are the large number of skeletal dysplasias and their phenotypic variability. The prevalence of skeletal dysplasias (excluding limb amputations) is estimated at 2.4 per 10,000 births. The most frequent are **thanatophoric dysplasia** *(Fig. 30 on page 52)*, **achondrogenesis**, **achondroplasia** *(Fig. 90 on page 115)*, and **osteogenesis imperfecta** *(Fig. 31 on page 53 Fig. 11 on page 30)*. They have been shown in thorax section *(2)*.
  - **Arthrogryposis** *(Fig. 91 on page 116, Fig. 92 on page 117, Fig. 93 on page 118)*: term that comprises a heterogeneous group of conditions that cause multiples joint contractures. Altered fetal movement is considered a contributor in pathogenesis. This restriction may be caused by neurologic or myogenic alterations, or secondary to extrinsic factors such as oligohydramnios. It usually affects the distal segments of the limbs. There may be muscle atrophy, flexion or extension contractures and joint ankyloses. The lesions can progress during pregnancy and after birth. In most cases they are bilateral but not necessarily symmetrical. The lower limbs are almost always affected, and more than half of the cases involve the upper limbs. The associated skeletal anomalies are scoliosis, clubfoot, clubhand, tarsal and carpal fusions and congenital hip luxation. Fractures can occur during labor.
  - **Amniotic band syndrome**: collection of fetal anomalies caused by the presence of amniotic fibrous tracts that entrap various fetal parts, mainly hands and feet. The reported incidence is 1:1,200 to 1,5000 live births. It should be suspected if there is an isolated fetal limb anomaly. The most common findings are limb amputations, acro-syndactyly, oligodactyly, clubfeet and craniofacial anomalies. The MRI appearance of amniotic bands is that of low T2 signal linear strands; they are reproduced on several sequences and planes as opposed to motion artifact in the amniotic fluid. Figures 32 and 81 show cases of this entity.
• **Soft tissue edema**: it can be focal as in Turner’s syndrome ([Fig. 94 on page 119](#)) or diffuse (hydrops fetalis) ([Fig. 95 on page ](#). Nowadays the most common etiology of hydrops is non-immune.

• **Lymphangiomas** ([Fig. 96 on page 121, Fig. 97 on page 122](#)): are lymphatic system alterations; they may consist of dilated lymphatic ducts or cystic hygroma with large cystic spaces in soft tissues.

### 6. OTHER: Table 9 on page 123

MRI failed to detect 164 anomalies. Pregnancy was voluntarily interrupted in 124 fetuses and 13 fetuses were stillborn.
<table>
<thead>
<tr>
<th>Fetal MRI anomalies</th>
<th>Number of fetuses</th>
</tr>
</thead>
<tbody>
<tr>
<td>Limbs and soft tissues</td>
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</tr>
<tr>
<td>Central nervous system</td>
<td>77</td>
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<tr>
<td>Head and neck</td>
<td>65</td>
</tr>
<tr>
<td>Thorax</td>
<td>57</td>
</tr>
<tr>
<td>Abdomen</td>
<td>47</td>
</tr>
<tr>
<td>Genitourinary</td>
<td>36</td>
</tr>
<tr>
<td>Other</td>
<td>33</td>
</tr>
<tr>
<td>Neural tube and spine</td>
<td>22</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td><strong>435</strong></td>
</tr>
</tbody>
</table>

**Table 1:** Fetal MRI anomalies found in the study with clinical and/or pathological correlation.

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<table>
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<tr>
<th>Central Nervous System anomalies</th>
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</tr>
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<tbody>
<tr>
<td>Hydrocephalus</td>
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<tr>
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<td>14</td>
</tr>
<tr>
<td>Chiari II</td>
<td>14</td>
</tr>
<tr>
<td>Cerebellar hypoplasia</td>
<td>9</td>
</tr>
<tr>
<td>Holoprosencephaly</td>
<td>7</td>
</tr>
<tr>
<td>Dandy-Walker malformation</td>
<td>2</td>
</tr>
<tr>
<td>Hydranencephaly</td>
<td>2</td>
</tr>
<tr>
<td>Cerebral tumor</td>
<td>1</td>
</tr>
<tr>
<td>Cerebral hemiatrophy</td>
<td>1</td>
</tr>
<tr>
<td>Glioependymal cyst</td>
<td>1</td>
</tr>
<tr>
<td>Intracranial hemorrhage</td>
<td>1</td>
</tr>
<tr>
<td>Lissencephaly</td>
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</tr>
<tr>
<td>Microcephaly</td>
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<tr>
<td>Macrocephaly</td>
<td>1</td>
</tr>
<tr>
<td>Rhombencephalosynapsis</td>
<td>1</td>
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<tr>
<td>Schizencephaly</td>
<td>1</td>
</tr>
<tr>
<td>Septo-optic dysplasia</td>
<td>1</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td><strong>77</strong></td>
</tr>
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</table>

**Table 2**

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Fig. 1: Alobar Holoprosencephaly. Fetus at 22 weeks’ gestation. (a-c) Fetal MRI axial and coronal T2-weighted images: (a) hypertelorism; (b,c) there is a small focus of brain within the anterior calvarium (asterisk) with a large dorsal cyst (arrows) pushing it forward. Images (d, e): gross pathology showing a small cerebrum with complete absence of interhemispheric fissure.

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Fig. 2: Semilobar Holoprosencephaly. Fetus at 22 weeks' gestation. (a-c) Fetal MRI coronal and sagittal T2-weighted images show (a) monoventricle (arrow) and fused basal nuclei (asterisk); (b) flattened nose (arrow); (c) cleft lip (arrow). (d-g) Anatomic specimen shows the same anomalies. Fetal MRI did not detect the polydactyly of the feet (arrow in d).

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**Fig. 3:** Semilobar Holoprosencephaly. Fetus at 21 weeks’ gestation. (a-c) Fetal MRI coronal and sagittal T2-weighted images showing fused basal nuclei (arrow in a), monoventricular system (asterisk in a) and absence of interhemispheric fissure. (b and c) show the associated facial anomalies (midline cleft lip, cleft palate, hypotelorism and flattened nose). (d-f) All the findings can be seen in the anatomical specimens.

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Fig. 4: Lobar holoprosencephaly. Fetus at 21 weeks’ gestation. (a-c) Fetal MRI coronal and sagittal T2-weighted images show continuity of the frontal lobes across the midline and facial anomalies. (d-f) Anatomic specimen showing the same intracranial anomaly (d) and facial anomalies: median cleft lip, hypotelorism and arhinia.

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Fig. 5: Agenesis of the corpus callosum. Fetus at 21 weeks’ gestation. (a,b) Fetal MRI coronal and sagittal T2-weighted images showing absence of the corpus callosum and cavum septum pellucidum; the frontal horns curve upwards (short arrow in a) and the upwardly displaced third ventricle (large arrow in a), characteristic findings for this entity, are well demonstrated. (b) Musculoskeletal anomalies were also found at MRI: multiple joint contractures in multiple areas, suggestive of arthrogryposis (it is a common association with agenesis of the corpus callosum). (c-e) Anatomic specimen of this fetus shows all of the findings described on MRI.

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Fig. 6: Agenesis of the corpus callosum. Fetus at 22 weeks' gestation (a) Fetal MRI coronal T2-weighted image shows absence of the corpus callosum and septum pellucidum, and prominent interhemispheric fissure (arrow) (b) Anatomic specimen demonstrates the absence of the callosal commissure with separation of the cerebral hemispheres (arrows).

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Fig. 7: Septooptic dysplasia. Fetus at 21 weeks' gestation. (a-c) Fetal MRI coronal T2-weighted images showing absence of the septum pellucidum (arrow) and fusion of the frontal horns of the lateral ventricles. (d) Anatomic specimen showing the absence of the septum pellucidum. Other anomalies were severe hypoplasia of the optical nerve and chiasm, severe hypoplasia of the neurohypophysis, and dysgenesis of the corpus callosum.

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**Fig. 8:** Cerebral hemiatrophy. Dyke-Davidoff-Masson syndrome. Fetus at 22 weeks' gestation. Cerebral hemiatrophy and facial lesions. (a-c) Fetal MRI coronal and sagittal T2-weighted images. There is marked dilatation of the right lateral ventricle (asterisks) and atrophy of all of the cerebral structures on the right side (arrow in a). Absence of right palate (circle in b and arrow in c). (d-f) Anatomic specimen showing the intracranial anomalies and right cleft lip and palate.

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**Fig. 9:** Microlissencephaly. Fetus at 34 weeks' gestation. (a) Fetal MRI sagittal T2-weighted image: microcephaly and small brain with smooth appearance of the brain surface. The same findings were found at birth (b), postnatal cranial radiography (c) and at brain MRI (d).

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Fig. 10: Schizencephaly. Fetus at 18 weeks’ gestation. (a, b) Fetal MRI sagittal and axial T2-weighted images. Note the ventriculomegaly and wide opening between the lateral ventricle and the cortical surface (arrow). (c,d) The same anatomical anomalies were found in the specimen.

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Fig. 11: Dandy-Walker Malformation (DWM). Fetus at 19 weeks' gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images. (a) DWM: Defect in the cerebellar vermix (arrow) and communication between the fourth ventricle and the cisterna magna. (b) Agenesis of corpus callosum (arrow) was also found. (c, d, f) Anatomic specimens showed DWM (c) and agenesis of corpus callosum (d). This fetus also suffered from Imperfect Osteogenesis, with femur (e) and rib fractures (f).

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Fig. 12: Rhombencephalosynapsis. Fetus at 20 weeks’ gestation. (a-c) Fetal MRI coronal and axial T2-weighted images show fusion of the cerebellar hemispheres without intervening vermis (arrow). (d-e) The fusion of the cerebellar hemispheres and absence of the vermis were confirmed in the anatomic specimen.

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Fig. 13: Brain tumor. Fetus at 35 weeks’ gestation. (a,b) Fetal MRI coronal and axial T2-weighted images showing macrocephaly secondary to a large heterogeneous mass in the left hemisphere (arrows), with significant hemorrhagic component (asterisk) and severe mass effect with midline shift and hydrocephalus. (c, d) The same findings were found at postnatal MRI. (e) Anatomic specimen after resection of the tumor: the final diagnosis was PNET (primitive neuroectodermal tumor).

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Fig. 14: Interhemispheric glioependymal cyst with agenesis of the corpus callosum and ventricular dilatation. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI coronal and axial T2-weighted images show ventricular dilatation (asterisk), an interhemispheric cyst (arrow in b) and absence of corpus callosum. (c, d) Anatomic specimen of this fetus shows all of the findings described at MRI.

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Fig. 15: Intracranial hemorrhage. Fetus at 25 weeks’ gestation. Ventriculomegaly was found at US. (a-d) Fetal MRI coronal and axial images showing ventriculomegaly and bilateral small hypointensities in the germinal matrix on T2-weighted images (short arrows in c); there is also a focal spot of T1-hyperintense and T2-hypointense signal in the frontoparietal region (dashed arrow in a and b) that corresponds to intraxial hemorrhage. (e, f) Anatomic specimen showing the intraparenchimal hemorrhage; there was another focus in the cerebellum that was not found at MRI (d).

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Fig. 16: Hydranencephaly. Congenital cytomegalovirus (CMV) infection. Fetus at 32 weeks' gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images showing the almost total destruction of the brain parenchyma, which is replaced by fluid cavities (asterisk); only the midline falx (short arrow in b) and basal nuclei (long arrow in b) are visualized. (c, d) Anatomic specimens showing the same anomalies. (e) Kidney histologic study found CMV nuclear inclusions as well as in other organs (arrow).

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<thead>
<tr>
<th>Head and Neck anomalies</th>
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<td>28</td>
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<td>Cleft palate</td>
<td>15</td>
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<tr>
<td>&quot;Nuchal fold&quot;</td>
<td>9</td>
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<td>Cystic hygroma</td>
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<td>Hypertelorism</td>
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<tr>
<td>Cystic lymphangioma</td>
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<tr>
<td>Nasal cavity tumor</td>
<td>1</td>
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<tr>
<td><strong>TOTAL</strong></td>
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</tr>
</tbody>
</table>

Table 3

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**Fig. 17:** Unilateral cleft lip. Fetus at 23 weeks’ gestation. (a) Fetal MRI axial T2-weighted image: the upper jaw is normal (white arrows) and the cleft lip can be seen with difficulty (black arrow); (b) sagittal image, the palate is intact (arrow). (c) Coronal image showing the lips (black arrows) and the cleft lip (red arrow). (d) Shows this child after birth.

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Fig. 18: Unilateral cleft lip and palate. Fetus at 21 weeks’ gestation. (a) Fetal MRI axial T2-weighted image showing the lesion of the upper jaw and lip (arrow). (b,c) Coronal images show the communication between the oral cavity and left nasal cavity (arrows). (d) Photo of this patient after birth. (e-g) The patient underwent reparative surgery; a small osseous defect is seen (arrow) at a CT performed 10 years later.

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**Fig. 19:** Bilateral cleft lip and palate. Fetus at 22 weeks’ gestation. (a) Fetal MRI sagittal T2-weighted image on the midline in which the palate is not seen (circle); (b,c) coronal images showing the communication between the oral cavity and the nasal cavities and the upper lip lesion (circle and arrows). (d) Photography after birth showing the cleft lip.

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Fig. 20: Epignathus. (a) Fetus at 22 weeks’ gestation. Fetal MRI sagittal T2-weighted image showing a complex mass (arrow) that appears to originate from the nasal cavities. (b) A follow-up MRI was performed 3 months later: the mass was separated from its pedicle and had moved (arrow). (c) Photo after birth showing the remaining mass in the right nasal cavity and (d) the biggest fragment found next to the newborn. Teratoma was the final diagnosis.

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Fig. 21: Epulis. Female fetus at 32 weeks’ gestation. (a,b) Fetal MRI sagittal and coronal T2-weighted images. There is a mass (arrow) protruding from the mouth. (c, d) Child at birth. (e) Histology of the lesion: gingival granular cell tumor composed of large pale cells with granular cytoplasm and background plexiform capillary network.

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**Fig. 22:** Goiter. Fetus at 25 week's gestation. (a,b) Fetal MRI coronal and sagittal T1-weighted images showing an enlarged fetal thyroid (arrows). (c, d) Normal fetal thyroid of another patient showing the significant difference in size. (e) Anatomic specimen showing the goiter (arrow) and thymus (asterisk).

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<table>
<thead>
<tr>
<th>Neural Tube and Spine anomalies</th>
<th>Number of fetuses</th>
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<td>Spina bifida</td>
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<tr>
<td>Encephalocele</td>
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<tr>
<td>Scoliosis</td>
<td>3</td>
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<td>Iniencephaly</td>
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<tr>
<td>Sacrococcygeal teratoma</td>
<td>1</td>
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</tbody>
</table>

**Table 4**

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Fig. 23: Encephalocele and vermian hypoplasia. Fetus at 21 weeks’ gestation. (a) Fetal MRI sagittal T2-weighted image. Small occipital cephalocele (arrow) and vermian hypoplasia (asterisk). (b-d) Gross pathology (b, c) showing evagination of a small part of the brain and leptomeninges through an occipital midline bone defect (arrow) and vermian hypoplasia (asterisk in d).

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**Fig. 24:** Closed spinal dysraphism with diastomyelia and tethered cord. (a, b) Fetal MRI coronal and sagittal T2-weighted images showing posterior lumbar spinal dysraphism (arrows). (c,d) Lateral and frontal spinal X-ray shows osseous defect and widening of the spine (white arrows) and heterotopic ossification behind L1, L2 and L3 posterior arcs (black arrow). (e) Subcutaneous mass at birth.

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Fig. 25: Open spinal dysraphism. Fetus at 20 weeks' gestation. (a,b) Fetal MRI axial and coronal T2-weighted images showing the bony defect (arrows). (c, d) Gross anatomy. The placode (arrow) is directly exposed to the environment.

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Fig. 26: Neural tube defect. Fetus at 17 weeks’ gestation. (a) Fetal MRI sagittal T2-weighted image showing hydrocephalus (asterisk), Chiari II (thin arrow) and a posterior defect at the lumbosacral spine (thick arrow). (b) Anatomic specimen showing the neural tube defect (arrow).

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**Fig. 27:** Open spina bifida. Fetus at 18 weeks’ gestation. (a,b) Fetal MRI axial and coronal T2-weighted images: hydrocephalus (asterisk), smaller than normal posterior fossa with caudal displacement of the vermis, brainstem, and fourth ventricle: Chiari II malformation (black arrow). Posterior spina bifida allowing communication between the spinal canal and amniotic sac (white arrows). (c, d) Arthrogryposis with club feet (black arrow) and important disorder of the spinal alignment and development.

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**Fig. 28:** Open spina bifida. Fetus at 17 weeks' gestation. (a, b) Fetal MRI axial and sagittal T2-weighted images showing hydrocephalus (asterisk), Chiari II malformation (white arrow) and open spinal dysraphism (black arrow). (c-d) Anatomic specimen showing the same spinal anomalies.

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Fig. 29: Sacrococcygeal teratoma with significant external component. Fetus at 36 weeks' gestation. (a) Fetal MRI sagittal image (obtained with long acquisition duration spin-echo technique) showing a large mass (arrows) arising from the sacrococcygeal region. (b) Afterbirth photograph. (c) Postnatal MRI sagittal T1-weighted image showing the cystic component of the mass (arrows). This study was performed in 1995, when HASTE sequences were not available in our centre.

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<table>
<thead>
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<th>Thoracic anomalies</th>
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<td>Diaphragmatic hernia</td>
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<tr>
<td>Congenital pulmonary airway malformation</td>
<td>9</td>
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<td>Cardiopathy</td>
<td>7</td>
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<td>Hydrothorax</td>
<td>7</td>
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<tr>
<td>Pulmonary sequestration</td>
<td>6</td>
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<tr>
<td>Pulmonary hypoplasia</td>
<td>5</td>
</tr>
<tr>
<td>Bronchial atresia</td>
<td>2</td>
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<tr>
<td>Thoracic lymphangioma</td>
<td>1</td>
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<tr>
<td><strong>TOTAL</strong></td>
<td><strong>57</strong></td>
</tr>
</tbody>
</table>

Table 5

© UDIAT-CD, Hospital Univeristari Parc Taulí, Sabadell.
**Fig. 30:** Thanataphoric dysplasia. Fetus at 18 weeks’ gestation: (a-c) Fetal MRI sagittal and coronal T2-weighted images; the thoracic cavity is narrow compared to the abdomen (arrows). The ribs are short, but this finding is difficult to assess at MRI. The final diagnosis was skeletal dysplasia with pulmonary hypoplasia. (d) Fetal radiograph after pregnancy termination showing the abnormal thoracic cavity and short ribs (arrow). Anatomic specimen (e,f) showed the same anomalies.

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**Fig. 31:** Osteogenesis Imperfecta. Fetus at 21 weeks’ gestation: (a) Fetal MRI coronal T2-weighted image: narrow thoracic cage and multiple rib fractures (arrows). There were more fractures in other bones (not shown). (b) Fetal radiograph showing multiple bone fractures. (c) Anatomic specimen shows the same findings.

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Fig. 32: Amniotic band syndrome. Fetus at 21 weeks’ gestation. (a,b) Fetal MRI coronal T2-weighted images showing thoracic deformity, severe scoliosis and hyperextension of the fetal head and flexion of the extremities. There is also pulmonary hypoplasia. (c) The anatomic specimen cannot be shown but the fetal radiograph shows the good correlation of MRI.

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**Fig. 33:** Iniencephaly. Fetus at 17 weeks’ gestation. (a,b) Fetal MRI sagittal T2-weighted images: severe spinal lesions, especially at the cervical level, and significant alteration of the morphology of the thoracic cavity causing marked pulmonary hypoplasia. Liver (arrow in a), heart (arrow in b), and there is hardly any space for the lungs. (c,d) Fetal radiograph and anatomic specimen showing this anomaly and the good correlation with the MRI.

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**Fig. 34:** Pulmonary hypoplasia due to thoracic deformity and diaphragmatic eventration. Fetus at 21 weeks’ gestation. (a-c) Fetal MRI axial, coronal, and sagittal T2-weighted images. The lungs are very small and the chest is deformed. Note the position of the spinal canal (arrow in a), which is not centered in the circumference of the thoracic curve. The left hemithorax is very small. The liver (arrows in a, b, and c) is very high. The thoracic deformity, the diaphragmatic eventration and the pulmonary hypoplasia are shown in the fetal radiograph (d) and anatomic specimen (e).

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Fig. 35: Left diaphragmatic hernia. Fetus at 22 weeks’ gestation. This fetus was easily diagnosed at US because the stomach is located within the thoracic cavity. (a) Fetal MRI coronal T2-weighted image shows rightward displacement of the heart (long arrow) and the stomach (short arrow) within the thoracic cavity; bowel loops are also depicted inside the thoracic cavity. (b) Anatomic specimen showing the good correlation with MRI.

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Fig. 36: Right diaphragmatic hernia. Fetus at 22 weeks’ gestation. (a) Fetal MRI coronal T2-weighted image. (b, c) Anatomic specimen. The heart is on the left side, almost in its usual position (discontinuous arrows in a and b) therefore it is difficult to assess the hernia by ultrasound. The liver (short arrows in a and b) is occupying the right hemithorax and there are bowel loops behind it (arrow in c). Note the pulmonary hypoplasia (long arrows in a and c).

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Fig. 37: Left diaphragmatic hernia. Fetus at 21 weeks' gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images. The stomach (long arrows) seems to be in the abdominal cavity. There is rightward displacement of the heart (short arrow in b) and pulmonary hypoplasia. (c) Anatomic specimen: the stomach is in the thoracic cavity. This finding did not correlate with the MR images; on MRI the stomach seemed to be located in the abdominal cavity (long arrow in b).

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Fig. 38: Left diaphragmatic hernia. Fetus at 22 weeks’ gestation. (a-c) Fetal MRI coronal and axial T2-weighted images: the left lobe of the liver (thick arrow in a) and some small bowel loops (blue arrow in b) are seen within the chest. Marked pulmonary hypoplasia (yellow arrows) and bilateral hydrothorax (long arrows) are shown. The same findings are shown in the anatomic specimen (d, e).

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**Fig. 39:** Diaphragmatic hernia, short neck and mega cisterna magna. Fetus at 24 weeks' gestation. (a-c) Fetal MRI coronal and sagittal T2-weighted images. Left diaphragmatic hernia is easily detected (long arrows); short neck (thick arrow in b); mega cisterna magna (short arrow in c). (d-f) Photographs of the anatomic specimen showing the anomalies.

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**Fig. 40:** Congenital pulmonary airway malformation (CPAM). Fetus at 24 weeks' gestation. (a-c) Fetal sagittal, coronal and axial T2-weighted images showing a polylobulated lesion (arrows) with septations that is occupying part of the right hemithorax. (d) In the plain chest radiography after birth the lesion is difficult to identify but the CT confirms the anomaly (e); the lesion appears to have decreased in size (compare c and e).

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**Fig. 41:** Congenital pulmonary airway malformation (CPAM). Fetus at 20 week’s gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images: CPAM occupies a large part of the right hemithorax (black arrow). There is leftward mediastinal shift and severe pulmonary hypoplasia of the right lung (white arrow in b). Follow-up ultrasound showed hidrothorax. The family opted for interrupting the pregnancy. Anatomic specimens (c, d) show the CPAM (black arrow) and pulmonary hypoplasia of the right lung (white arrow).

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Fig. 42: Congenital pulmonary airway malformation (CPAM). Fetus at 21 weeks' gestation. (a-c) Fetal MRI sagittal, coronal and axial T2-weighted images show cystic structures of middle size (arrows) in the right lung base. After birth the cysts increased in size and therefore they underwent surgical excision. (d) CT prior to surgery and (e) surgical piece of the same patient.

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**Fig. 43**: Extralobar sequestration. Fetus at 23 weeks’ gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images. A triangular-shaped lesion (arrows) is seen in the left hemithorax base; it corresponds to pulmonary sequestration. (c) CT after birth showing the same lesions and (d, e) surgical pieces.

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Fig. 44: Extrapulmonary sequestration. Twin pregnancy. (a, b) Fetal MRI images: a lesion is seen in the left lung base of one of the fetus (arrows). (c, d) CT after birth shows the same lesion with vessels and systemic venous drainage (arrows). In this case the photograph of the anatomic specimen is not available.

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**Fig. 45:** Extralobar sequestration and Intestinal duplication. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images. The short arrows show gastric duplication, the stomach is marked by long arrows, and the pulmonary sequestration by thick arrows. The first US study only detected intestinal duplication. (c) Anatomical piece showing the same anomalies.

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**Fig. 46:** Segmental bronchial atresia. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI coronal T2-weighted images showing a pulmonary lesion (arrows) that occupies a large part of the upper left lung lobe. (c) Postnatal radiography and (d) coronal CT image show the same lesion. The patient underwent surgery; the histological study (not shown) confirmed the diagnosis.

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Fig. 47: Segmental bronchial atresia. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI coronal and sagittal T2-weighted images: an hyperintense lesion (arrows) can be seen in the upper right lobe of the lung. (c, d) Postnatal CT shows the good morphologic correlation of the lesion between the studies. (e) Anatomic pieces of the anomaly after resection.

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**Fig. 48:** Hydrothorax, complex cardiopathy and agenesis of the corpus callosum. Fetus at 22 weeks' gestation. (a-c) Fetal MRI T2-weighted images showing severe pulmonary hypoplasia and hydrothorax (arrows in a, b). Note also agensis of the corpus callosum (a), vermian hypoplasia (b) and significant ascites and edema of the soft tissues (arrow in c). (d) Fetal radiograph and (e) Anatomic specimen showing the anomalies.

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Fig. 49: Meckel-Gruber syndrome (MGS). Pulmonary hypoplasia due to absence of amniotic fluid. Fetus at 24 weeks' gestation. (a) Fetal MRI coronal image showing severe oligohydramnios and bilateral pulmonary hypoplasia (short arrows). Enlargement of both kidneys with multiple cysts (long arrow). (b) Anatomic specimen shows the same anomalies. Note also an encephalocele (thick arrow) and the hypoplastic right lower limb that was not seen with MRI.

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**Fig. 50:** Obstruction of the lower urinary tract. Fetus at 17 weeks’ gestation. (a) Fetal MRI coronal image and (b) Anatomic specimen showing the pulmonary hypoplasia (short arrows) resulting from oligohydramnios caused by obstruction of the lower urinary tract due to urethral atresia with megacystis (long arrow).

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**Fig. 51:** Cardiopathy. Endocardial fibroelastosis. Fetus at 25 weeks' gestation. (a-c) Fetal MRI T2-weighted images show significant cardiomegaly (arrow) and pulmonary hypoplasia. (d-f) Histological examination established the diagnosis of endocardial fibroelastosis.

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**Fig. 52:** Cardiopathy and holoprosencephaly. Fetus at 22 weeks’ gestation. (a) Fetal MRI coronal T2-weighted coronal image shows prominent right heart chambers (arrow), and holoprosencephaly (short arrow). (b) Anatomic specimen showing the cardiomegaly.

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**Fig. 53:** Heterotaxy syndrome and complex cardiopathy. Fetus at 20 weeks' gestation. (a, b) Fetal MRI coronal T2-weighted images. The liver (short arrows in a) and the stomach (long arrow) are malpositioned in the abdomen. Heart (black arrow in a). (c) Anatomic specimen showing the same findings (arrows).

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**Table 6**

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**Fig. 54:** Duodenal diaphragm and cloacal malformation. Fetus at 31 weeks’ gestation. (a, b) Fetal MRI coronal and sagittal T2-weighted images. Gastric and duodenal distension (arrows in a). Cloacal anomaly is seen in image (b): bladder (short arrow) and vagina (long arrow). (c) X-ray after birth shows gastric and duodenal distension. (d) Surgical intervention showed duodenal diaphragm. (e) Patient’s photograph after birth showing the cloacal malformation (anal atresia). (f) Selective study of the cloacal malformation: arrows show bladder (B), urethra (U) and vagina (V); there is also vesicoureteral reflux (VUR).

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Fig. 55: Duodenal obstruction due to annular pancreas. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI coronal and axial T2-weighted images. Gastric (long arrow) and duodenal (short arrow) dilatation. There is also polyhydramnios. (c) Postnatal radiograph showing duodenal obstruction. No air is seen distally in the intestine. (d) Surgical intervention demonstrated annular pancreas as the cause of the duodenal obstruction.

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**Fig. 56:** Duodenal stenosis due to Ladd's band. Fetus at 32 weeks’ gestation. (a) Coronal T2-weighted images of the fetus. The duodenum is greatly dilated until the third portion (long arrow), the stomach (short arrow) seems normal. The volume of amniotic fluid is normal. The large bowel is in the left abdomen. (b) X-ray with oral contrast after birth and (c, d) photos of surgical intervention confirm this anomaly. Meckel's diverticulum (arrow in c), undetected at US and MR, was present.

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**Fig. 57:** Jejunal atresia. Fetus at 25 weeks’ gestation. (a) Coronal T2-weighted and (b) coronal T1-weighted sequences of the fetal MRI showing dilatation of some proximal small bowel loops with meconial content (arrows). (c, d) Postnatal X-ray and barium study showing dilatation of the jejunal loops and microcolon (arrows). (e) Postnatal photograph shows abdominal distension. (f) Surgical intervention demonstrated jejunal atresia.

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Fig. 58: Multiple jejunal atresia. Fetus at 22 weeks’ gestation. Fetal MRI Coronal (a) T2-weighted and (b) T1-weighted images showing jejunal distension with meconial content inside (arrows). (c) Postnatal X-ray shows jejunal occlusion (arrows). (d) Surgical intervention demonstrated multiples jejunal atresia (arrows). The patient died a few days after the surgery.

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Fig. 59: Cecal atresia. Fetus at 24 weeks’ gestation. Fetal MRI coronal (a) T2-weighted and (b) T1-weighted images showing distension of an intestinal segment in the right flank, with meconium inside (arrows). (c) Postnatal X-ray and (d) surgical intervention showed cecal atresia and the good correlation with fetal MRI.

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Fig. 60: Intestinal duplication. Fetus at 24 weeks’ gestation. (a, b) Fetal MRI coronal and sagittal T2-weighted images; an hyperintense structure (arrows) is seen above the bladder. (c) X-ray after birth showing intestinal occlusion; (d, e) surgery discovered intestinal duplication and volvulus.

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**Fig. 61:** Ovarian cyst. Fetus at 35 weeks’ gestation. (a-c) Fetal MRI T2-weighted images showing a cystic mass in the right abdomen (long arrow) above the bladder (short arrow). (d) Postnatal US and (e) surgical intervention demonstrated this ovarian cyst.

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**Fig. 62:** Hepatic hemangioendothelioma. Fetus at 35 weeks’ gestation. (a-c) Coronal, axial and sagittal T2-weighted images showing a mass in the left hypochondrium between the liver, spleen and stomach. (d) Postnatal CT showed the mass (arrows) with heterogeneous and peripheral contrast enhancement, suggestive of vascular tumor. (e) Surgical intervention confirmed the hepatic origin of the tumor (arrow).

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Fig. 63: Hepatic cyst. Fetus at 17 weeks’ gestation. This fetus with agenesis of one kidney and dysplasia of the other also had a cyst in the upper part of the abdomen. (a) Fetal MRI sagittal T2-weighted image showing the cyst (arrow). (b, c) The hepatic cyst can be seen in the pathological specimen.

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Fig. 64: Ascites. Fetus at 19 weeks’ gestation. Polymalformation. (a-c) Fetal MRI T2-weighted images showing cleft lip (long arrow in a), vermian hypoplasia (long arrow in b), ascites (short arrows in a-c) and skeletal deformities characteristic of osteogenesis imperfecta. (d) Fetal X-ray and (e) Anatomic specimen showing the same skeletal anomalies.

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**Fig. 65:** Gastroschisis. Fetus at 20 weeks’ gestation. (a-c) Fetal MRI T2-weighted images showing the bowel loops (black arrow) outside of the abdominal cavity. There is a right lateral abdominal wall defect, through which the loops herniate (small arrows in a and c). (d) Postnatal photograph shows the anomaly and the good morphologic correlation with MRI.

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Fig. 66: Gastrochisis. Fetus at 22 weeks’ gestation. (a, b) Fetal MRI T2-weighted images: both in the sagittal (a) and axial (b) views, we can see the bowel loops outside of the abdominal cavity (arrow in a). There is no peritoneal lining and the umbilical cord is inserted in the correct position (arrow in b). (c, d) Photographs of the patient after birth and after the surgical reparation.

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Fig. 67: Omphalocele. Fetus at 21 weeks’ gestation. (a,b) Fetal MRI sagittal and axial T2-weighted images show the bowel loops outside the abdominal cavity (long arrow) lined with peritoneal membranes (small arrows) and the umbilical cord is inserted in the omphalocele (thick arrow). (c) Volumetric SS-RARE images are useful to identify the umbilical cord (arrow). (d) Postnatal photograph.

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Fig. 68: Omphalocele. Fetus at 18 weeks’ gestation. (a-c) Fetal MRI T2-weighted images showing an omphalocele. Part of the liver (arrows) is outside of the abdominal cavity. (d) Postnatal photograph showing the peritoneal membrane covering the liver; the umbilical cord is also involved.

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**Fig. 69:** Inguinal hernia. Fetus at 30 weeks’ gestation. (a, b) Fetal MRI sagittal and axial T2-weighted images: we can observe scrotal distension (long arrow in a) and the testes (short arrows in a and c), the inguinal canal appears exceedingly open (arrow in b). After birth (d) he underwent surgery. The hernia contained epiplon.

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Table 7

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Fig. 70: Cystic renal dysplasia. Fetus at 22 weeks’ gestation. (a-c) Fetal MRI coronal T2-weighted images showing cystic structures occupying significant part of the fetal abdomen. The morphology of the right kidney and the bladder (short arrow in b) are normal. (c) Postnatal MRI showed the same findings. Because its significant size and compression of the contralateral urinary tract, this kidney was surgically removed (d).

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Fig. 71: Ambiguous genitalia. Fetus at 28 weeks’ gestation. (a-c) Fetal MRI axial T2-weighted images showing the external genitalia. It is not clear if there is male genitalia with micropenis or female with hypertrophied clitoris (arrows). (d, e) Postnatal photographs: the patient was male with micropenis and hypospadias; this genital morphology is very similar to the findings at MRI.

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Fig. 72: Ambiguous genitalia. Fetus at 25 weeks’ gestation. (a, b) Fetal MRI axial T2-weighted images of the fetal external genitalia: they are ambiguous and seem female with slightly hypertrophic clitoris (arrows); bladder is also seen (asterisk). Amniocentesis revealed that the fetus was a male. (c) Postnatal US showing the testicles (arrows) in the inguinal canal. (d) Photograph of the same patient showing micropenis (arrow), extreme hypospadias and rudimentary scrotum.

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Fig. 73: Bilateral renal cystic dysplasia. Fetus at 28 weeks’ gestation. (a, b) Fetal MRI images. There is severe oligohydramnios, therefore fetal evaluation is more difficult. Note the enlarged kidneys (thin arrows) with multiple cysts and mild dilatation of the pelvis (thick arrows); the bladder is empty. Pregnancy was interrupted. (c) Photograph of the fetus and (d, e) anatomic specimen showing the enlarged kidneys with multiple cysts, predominantly cortical.

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**Fig. 74:** Meckel-Gruber syndrome. (Same case as fig.49 in thorax section). Meckel-Gruber syndrome is a lethal autosomal recessive inherited syndrome with a 25% risk of recurrence. Polycystic kidneys, polydactylism, and encephalocele are the characteristic findings. (a, b) Fetal MRI images and (c, d) anatomic specimen. Some years later the woman got pregnant again and the fetus had the same syndrome.

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Fig. 75: Bilateral renal aplasia (aplastic dysplasia). Fetus at 19 weeks' gestation. (a, b) Fetal MRI sagittal and coronal T2-weighted images. There is severe oligohydramnios (arrows), that makes this fetus difficult to evaluate with US and MR. The fetal kidneys were not identified and the bladder was apparently empty. Termination of pregnancy was advised. (c, d) Anatomic specimen: there is facial and limb deformities (Potter sequence), the kidneys were rudimentary (renal aplasia) with ureteral and bladder agenesis. The fetus also had esophageal atresia, sigmoid atresia and imperforate anus; these anomalies were not detected at MRI.

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**Fig. 76:** Prune Belly syndrome. Same case as fig. 50 in thorax section. Fetus at 17 weeks' gestation. (a, b) Fetal MRI T2 weighted images showing great dilatation of the bladder (asterisk) causing distension of the abdomen and atrophy of the musculature of the abdominal wall. This fetus had Down’s syndrome and hydrocephalus (arrow). Esophageal atresia and annular pancreas were undetected at both US and MR. (c, d) Pathological specimen of this fetus.

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**Fig. 77:** Urachal cyst. Fetus at 16 weeks’ gestation. Twin pregnancy. One of the fetuses has a severe anomaly in the abdomen; the arrow in (a) indicates the bladder, the urachal cyst is shown (asterisk). (b, c) Pathological specimen of this fetus showing these anomalies. This fetus had urogenital tract anomalies and imperforate anus.

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Table 8

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**Fig. 78:** Total aplasia of the hand and partial of the forearm (transverse hemimelia). Fetus at 21 weeks’ gestation. (a) Thick-slab T2-weighted images showing absence of the distal half of one upper limb (arrow). (b) Radiograph after birth shows the same anomaly. Currently we do not dispose of the clinical image.

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**Fig. 79:** Hypoplastic or absent fibula. Fetus at 18 weeks’ gestation. (a, b) Fetal MRI images: note the shortening and deformity (arrows) of the right lower limb. The absence of the fibula and the curvature of the tibia are difficult to evaluate at MRI. (c) Fetal radiograph and (d) anatomic specimen showing the lesion. This anomaly is usually sporadic and is associated with limb shortening, tibial hypoplasia and feet malformation.

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**Fig. 80:** Acheiria. Fetus at 21 weeks’ gestation. (a, b) Fetal MRI images showing this anomaly (arrows). The absence of the fetal hand may be caused by amniotic band syndrome or be part of other syndromes such as Cornelia de Lange syndrome and fetal hydantoin syndrome. (c) Anatomic specimen showing this anomaly (arrow). Note the good correlation with the MR images.

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**Fig. 81:** Foot amputation due to an amniotic band. Fetus at 34 weeks' gestation. (a) Fetal MRI image showing this anomaly (arrow). Presently there is no clinical photograph of the anomaly but the postnatal radiograph is presented and demonstrates this anomaly and the good correlation with MRI.

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**Fig. 82:** Ectrodactyly. Fetus at 23 weeks’ gestation. (a) Fetal MRI thick slab T2-weighted image showing an abnormal morphology of the right hand (arrow). There were no other fetal anomalies. (b) Fetal radiograph and anatomic specimen (c-e) showing this anomaly in both hands and syndactyly in the left foot (arrow in e) that was not detected at MRI.

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Fig. 83: Bilateral Clubfeet. Fetus at 21 weeks' gestation. (a) Fetal MRI T2-weighted image: misaligned feet (arrows). It is usually isolated (as in this case) but sometimes there are other conditions. (b) Photograph of the anomaly at birth.

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Fig. 84: Left clubfoot. Fetus at 22 weeks' gestation. (a, b) Fetal MRI thick-slab T2-weighted sequence is the best to evaluate the morphology of the limbs. The thickness is 80 mm; it enables a good visualization of the lower limbs in this fetus. There is left clubfoot (arrows). Photographs after birth confirm the diagnosis.

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**Fig. 85:** Bilateral clubfeet. Fetus at 21 weeks’ gestation. (a) Fetal MRI thick-slab T2-weighted image showing bilateral clubfeet (arrows); note the good correlation with the postnatal photographs (b, c).

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Fig. 86: Fibular aplasia and feet malformation. Fetus at 35 weeks’ gestation. (a, b) Fetal MRI images: deformity of the lower limbs and positional anomaly of the feet (arrows). The toes seem abnormal (short arrows). (c) Postnatal photograph shows the same anomalies. Fibular aplasia (arrow in d) was not seen at MRI.

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**Fig. 87:** Congenital absence of the radius with bilateral clubhand; radial hemimelia. Fetus at 21 weeks’ gestation. (a-c) Fetal MRI images: abnormal shape and shortening of the upper limbs (arrows); radial aplasia was not seen in this study, (d, e) Postnatal radiographs and (f) photograph of the patient showing this anomaly. This patient also had hypospadias that was not detected neither at US nor MRI.

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Fig. 88: Genu recurvatum. Fetus at 18 weeks’ gestation. (a, b) Fetal MRI T2-weighted images: hyperextension of the knees (arrows). Most cases are bilateral; it may be isolated or associated with other pathologies. This fetus also had fetal growth restriction, oligohydramnios and renal and CNS anomalies. (c, d) Fetal radiograph and anatomic specimen.

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Fig. 89: Facio-genito-popliteal syndrome (popliteal pterygium syndrome). Fetus at 22 weeks' gestation. (a-c) Fetal MRI: cleft lip and palate (arrows in a and b) and popliteal pterygium (arrow in c). These anomalies are seen in the anatomic specimen (d-g). Toenails anomalies, characteristic in this syndrome, were not seen at MRI (arrow in g).

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**Fig. 90:** Achondroplasia. Fetus at 34 weeks’ gestation. (a-c) Fetal MRI T2-weighted images: disproportionate large cranium to the thorax and abdomen (arrows in a), prominent forehead (arrow in b) and short lower limbs. Both US and MRI suspected a skeletal dysplasia. (d, e) Postnatal photographs of the patient show the typical phenotype of achondroplasia.

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**Fig. 91:** Arthrogryposis. Fetus at 22 weeks’ gestation. (a) Fetal MRI sagittal T2-weighted image: lower limbs extension and elbow flexion (arrows). This position did not change during the whole study. This fetus also had hydrocephalus (asterisk). (d) Anatomic specimen showing the position of the limbs.

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Fig. 92: Arthrogryposis. Fetus at 20 weeks’ gestation. (a, b) Fetal MRI images and (c, d) Anatomic specimen: all limbs are flexed.

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**Fig. 93**: Arthrogryposis. Amniotic band syndrome. Fetus at 21 weeks’ gestation (same case as fig 32 in thorax section). (a) Fetal MRI thick-slab T2-weighted image: significant scoliosis; fetal limbs are flexed permanently. Amniotic bands are seen as lineal structures inside the uterus (arrow). Pregnancy was terminated; the fetus was covered in multiple amniotic bands. (b) Fetal radiograph: significant scoliosis and arthrogryposis.

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Fig. 94: Turner’s syndrome. Fetus at 22 weeks’ gestation. (a-c) Fetal MRI T2-weighted images: nuchal septated edema (arrow); (d) dorsal edema in the hand. Karyotype analysis confirmed Turner's syndrome. Fetal presentation of this syndrome has poor prognosis. (e, f) Nuchal fold and dorsal edema of hands and feet.

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**Fig. 98**: Hydrops fetalis. (a-c). Fetal MRI T2-weighted images: soft tissue edema (small arrows in a and b), pleural effusion (long arrow in c) and pulmonary hypoplasia (short arrow in c). (d, e, f) Fetal radiograph and anatomic specimen showing diffuse edema. The etiology in this case is unknown.

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**Fig. 96:** Thoracic lymphangioma. Fetus at 36 weeks' gestation. (a, b) Fetal MRI T2-weighted images: cystic mass in the left axilla (arrow). Postnatal studies confirmed the diagnosis. (c) Postnatal external appearance of the mass.

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Fig. 97: Thoracoabdominal wall lymphangioma. Fetus at 21 weeks' gestation. (a-c) Fetal MRI T2-weighted images: cystic mass in the thoracoabdominal wall (arrows). (c) Good correlation of this anomaly with the fetal MRI.

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<table>
<thead>
<tr>
<th>Other anomalies</th>
<th>Number of fetuses</th>
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<td>Oligohydramnios</td>
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<td>Polyhydramnios</td>
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<td>Abnormal placenta</td>
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<tr>
<td><strong>TOTAL</strong></td>
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Table 9

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Conclusion

The findings of fetal anomalies on MRI correlate well with the clinical and pathologic findings. MRI cannot detect all fetal anomalies, but it can detect those with poor prognoses that lead to the decision to voluntarily interrupt pregnancy. MRI is useful for clinical and surgical decision making and helps patients understand the anomalies.

Correlation of MRI findings with clinical and pathologic studies is essential for the radiologist in order to learn and improve about fetal pathology.

The radiologist should be part of the fetal pathology committees and participate actively in order to provide the most of the fetal MRI in the diagnosis, management and research of fetal pathology.
References