Radiographic features comparison of Patau and Edwards Syndromes with case reports illustration.

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Learning objectives

Compare the radiologic findings between two rare chromosomal syndromes that are among the differential diagnoses of each other.
Background

Rare chromosomal syndromes, first described in 1960 (2, 3, 4).

The trisomy 13 has a prevalence of 1 case per 5000-12000 births (2, 3), the third most common multiple malformation (5), while trisomy 18 is estimated to 1:6000-1:8000 (6; 7, 8, 9, 10) with a larger gestational prevalence (1:2500-1:2600), because of high rates of fetal loss and pregnancy interruption after prenatal diagnosis (6), being the second most common, trisomy being the two behind 21 trisomy (5, 6, 7, 8, 9, 11).

In cases of 13 trisomy during gestation until 12 weeks, about 49% end in miscarriage or stillbirth, and between 18 weeks and 40 weeks were 42%. From week 24, about 35% (13). Only 33% of fetuses can be born with life (12). When is a 18 trisomy, during pregnancy until 12 weeks, about 72% end in miscarriage or stillbirth, and between 18 weeks and 40 weeks were 65%. From week 24 is about 59% (13).

Both have approximately one recurrent every 100 pregnancies (5, 6) and has its increasing prevalence with increasing age (2, 6, 14). Nearly all live births die in the first year of life, showing the two syndromes, predominance of girls (15).

In most cases of Patau syndrome, the extra 13 chromosome for the trisomy (14) is originated by the mother (16), with some reports whose hypothesis is to be a non-disjunction of this chromosome (17, 18). However, in 80% of cases, mutation occurs with no maternal separation in meiosis (19). Mosaicism presents rarely, occurring in only 5% of patients (16, 20, 21), with variable (22) and poorly understood phenotype (16).

In the Edwards syndrome, the chromosomal disorder occurs due to the presence of an extra 18 chromosome (15), mosaic trisomy or partial 18q trisomy (6, 23). The changes may be caused by abnormal expression of important genes for 18 chromosome development (24). Most cases are caused by a phenomenon of non-disjunction during the second meiotic division of mother oogenesis (6). Translocations and mosaicism are less frequent, being observed in only 10% of cases (25, 26), and also being milder forms of the syndrome (23).

Patau syndrome is characterized by multiple and various malformations, being the major abnormalities severe mental retardation, hypotonia, skeletal malformations and midline facial defects, holoprosencephaly - alobar form being the most common (27), heart defects, omphalocele and polydactyly, presenting a very short survival (2, 16, 28), rarely achieving childhood (29, 30). Less associated anomalies, such as polyhydramnios,
oligohydramnios, intrauterine growth retardation, single umbilical artery, eye defects, such as congenital glaucoma (31) and polycystic kidneys may contribute to its clinical course (29, 32). Some skin defects have been reported as defects in the scalp, glabellar strains, deep palm creases, convex soles, rocker-bottom and multiple hemangiomas (14).

Its diagnosis is still suspected in prenatal exams, in the morphological abnormalities detected by ultrasound (29, 33) in about 90% of cases (34), however, the diagnosis is made at birth, with the karyotype evaluation (35) or a cordocetese and / or amniocentesis during pregnancy (36), to differentiate it from its main differential diagnosis: Meckel-Gruber syndrome (27).

The 18 trisomy anomalies involve the urogenital tract, such as cryptorchidism (27, 37), cardiovascular, craniofacial and central nervous system (6, 23). The pattern recognizable syndrome consists of major and minor anomalies (6), being the most common major malformations and are the most frequent, the cardiac anomalies - ventricular septal defect - and kidney - horseshoe kidneys (6, 38). Other major anomalies consist in intrauterine growth retardation, postnatal, psychomotor and cognitive growth deficiency (6).

The minor anomalies consist of head anomalies - strawberry-shaped head, the absence of the corpus callosum (27, 37), and micrognathia, dolichocephaly (43), cleft lip or palate (37) - and skeletal - small nails, underdeveloped thumbs, short sternum (6, 39), incomplete ossification of the clavicle, equinus varus foot, radial aplasia (37) and the clenched fist with overlapping fingers (27, 40), especially the second on the third finger on the association of the fifth finger clinodactyly (37). Hands anomalies are highly suggestive of Edwards Syndrome during prenatal examinations (27, 41, 42), and are, usually, bilateral (37).

The need for differential diagnosis of syndromes like Roberts syndrome, Smith-Lemli-Opitz syndrome, osteogenesis imperfecta, 13 trisomy, Cockayne Syndrome and Werdnig-Hoffmann disease should be considered (43, 44).

13 trisomy, unlike 18 trisomy, presents most frequently malformations evident on physical examination, which are characteristic pattern of multiple congenital anomalies. In general, the combination of orofacial clefts (cleft palate), microphthalmia and / or anophthalmia and post-axial polydactyly allows members of its recognition. This characteristic triad is observed in 60-70% of cases (15). Unlike Down and Edwards syndrome, the measure of the pelvic bone angle in the second trimester fetus has no value in the diagnosis of 13 trisomy (45).

**MORPHOLOGICAL ULTRASOUND FINDINGS ACCORDING TO MEDICAL LITERATURE**

<table>
<thead>
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Intrauterine growth retardation (15;29;32)

Holoprosencephaly - alobar form is the most common (15, 27)

Microcephaly (15)

Sloping forehead (15)

Micrognathia (15)

Ophthalmologic abnormalities (15)

Cleft lip or palate (15)

Short neck (15)

Apnea episodes (15)

Ventricular septal defect (6, 38) - communication between the ventricles (6, 15, 27, 37, 40)

Diaphragmatic hernia (46)

Omphalocele (27, 56)

Polycystic kidneys (15, 29, 32)

Single umbilical artery (29, 32)

Polydactyly (15, 29, 30)

Overlapping fingers (15)

Congenital clubfoot (32)

Oligohydramnios or polyhydramnios (29, 32)

Intrauterine growth retardation (6;15;27;37)

Corpus callosum absence (27, 37)

Microcephaly (15)

Strawberry shaped head (27; 37)

Micrognathia (15)

Choroid plexus cysts (27)

Dysmorphic facial skeleton - cleft lip or palate (6, 27, 37)

Esophageal atresia (38)

Ventricular septal defect (6, 38), communication between the ventricles and communication between the atria (6, 15, 27, 37, 40)

Cryptorchidism (6, 15, 27, 37)

Omphalocele (27, 56)

Horseshoe kidneys (6, 15, 37, 38)

Single umbilical artery (29, 32)

Fist with overlapping fingers (15, 27, 40), especially the second on the third finger on the association of the fifth finger clinodactyly (37)

Radial aplasia (1, 32, 33)
The main structures analyzed the ultrasound of the first trimester are the fetal nuchal translucency thickness, fetal heart rate, fetal nasal bone, fetal tricuspid regurgitation, ductus venous flow, fetal crown-rump length, fetal trunk and head volume, fetal frontomaxillary facial angle, gestational sac volume and umbilical cord diameter (22).

The positive predictive value of fetal nuchal translucency (cut-off of 2.5 mm) increased as a screening test for the detection of fetus chromosomal abnormalities is 14.8% for genetic malformations in general, with 2.45% for Patau Syndrome, which was confirmed after genetic testing corroborative (47). However, the most common ultrasonographic changes, regardless of the fetal echocardiography, are holoprosencephaly and facial defects in 13 trisomy (5).

On examination between 11 and 13 weeks, the increased nuchal translucency measurement and fetal heart rate, associated with assessment for holoprosencephaly and omphalocele can identify 90% of fetuses with 13 trisomy (48). Between 16-22 weeks, all cases presented any changes to the ultrasonographic examination, morphological or not, as the polyhydramnios and intrauterine growth retardation (49). However, omphalocele without holoprosencephaly is most common in Edwards Syndrome, followed by 13 trisomy (36, 56).

Ultrasound has a sensitivity of almost 100% for the visualization of structural defects (45), for example, increased nuchal thickness, intrauterine growth retardation, choroid plexus cyst (3), cerebellar hypoplasia, overlapping fingers, diaphragmatic hernia and congenital heart defects - communication between the atria and communication between the ventricles and single umbilical artery in 80% of cases (2, 45, 50, 51). Choroid plexus cysts after 22 weeks of pregnancy is a marker of 18 trisomy, as well as the Dandy-Walker malformation (27). Aspects for Edwards syndrome are detectable in 80% of affected fetuses in the second trimester (58).

Currently the majority of cases of 18 trisomy is suspected on prenatal (40), based on tracking by maternal age, screening of maternal serum marker or the detection of ultrasonographic abnormalities (6, 40, 27, 37). The diagnosis is made by analysis of fetal chromosomal map using material obtained at amniocentesis, chorionic villus sampling or cordocentesis (27).

Cardiac abnormalities are common in Patau Syndrome, with studies in the literature citing it as the most recurrent alteration followed by abnormalities in the central nervous system (50, 52) and facial anomalies (53), being diagnosed more commonly in the second
and third gestation trimesters, in association, in most cases with cystic hygroma, nuchal edema, fetal hydrops, omphalocele, diaphragmatic hernia, limbs abnormalities, such as clubfoot (32), genito-urinary abnormality and polydactyly (46).

There are reports with 100% sensitivity and 98.9% specificity for diagnosing prenatal genetic noninvasive 13 trisomy by sequencing of maternal DNA (54). In ultrasound, evaluation of fetal face (including ears) and the extremities (including hands and feet), with extensive use of fetal echocardiography increases the detection method sensitivity of the pathology (29).

In cases of free trisomy of 18 chromosome there’s no indication to perform cytogenetic evaluation of the parents, because this abnormality, as already explained, is due to a phenomenon of nondisjunction during gametogenesis. However, the accurate diagnosis of these syndromes is essential for assessment, clinical management and appropriate genetic counseling (15).

The prenatal screening and maternal age have dramatic effects on the births prevalence of these pregnancies. On the other hand, the survival of these diseases remains practically unchanged with most dying in the neonatal period (55).

CASE REPORTS

CASE 1

Female patient, 1 day old, caucasian, brazilian, born in Santos (SP), living in Praia Grande. Born by cesarean with presence of meconium, after 39 weeks of pregnancy, Apgar on first minute scoring 1 and 7 after 5 minutes, weighing 1600g, 24 cm of cephalic perimeter and 43 cm in length, hypotonic, heart rate of 70 beats per minute and irregular breathing. After emergency procedures, the heart rate rase up to 120 beats per minute. Physical examination showed tachypnea, microcephaly, absence of nose, extensive cleft palate, hypophonetic heart sounds, bilateral crackels, innocent abdomen without visceromegalies, ambiguous genitalia, umbilical cord with 1 artery and 1 vein, skin with vernix stained with meconium, and polydactyly in both hands. Readily transferred to the neonatal ICU, surviving for 1 month, until her death by respiratory failure.

Mother, 46 years old, healthy, public servant, drink alcohol socially (not ingested during pregnancy), having no previous pregnancy or birth complications. Nine held consultations in prenatal, complications as having minor bleeding in the 1st trimester of pregnancy and abnormal vaginal discharge on the 7th month of pregnancy being treated with Nystatin. Discovered malformations of the fetus in the 1st trimester of pregnancy by morphological ultrasound. Although judicial authorization, not opted for abortion. Serology for syphilis,
HIV, toxoplasmosis, hepatitis B and C showing negative IgG and IgM. For rubella and cytomegalovirus positive IgG but negative IgM. Father with 51 years, standalone, without vices, ex-smoker. There is no consanguinity between the parents and grandparents, both paternal and maternal.

At the 1st morphological ultrasound (Figures 1 to 5), performed at 14 weeks of gestation, it was found a normal nuchal translucency (1.1 mm), but no nasal bone, no characterization of the central nervous system midline anatomy, polydactyly in both hands, reverse wave A of the ductus venous Doppler, placental edge overlying the internal cervical canal and the umbilical cord with single umbilical artery, indicating research of fetal karyotype and fetal echocardiography.

The 2nd morphological ultrasound (Figures 6 to 13), performed at 21 weeks of gestation, characterized holoprosencephaly, microcephaly, facial dysmorphism (bilateral cleft lip, nose agenesis and hypotelorism), polydactyly, single umbilical artery, cardiopathy (ventricular septal defect and left heart chambers hypoplasia), intrauterine growth retardation and probable esophageal atresia.

With 22 weeks and 4 days of gestation, fetal echocardiographic doppler (Figures 14 to 17) was performed, confirming enlarged left ventricle and reduced right ventricle dimensions, interventricular communication bad alignment type with the aorta riding over the ventricular septal around 40% and not identified connection between the right ventricle and the pulmonary artery.

At birth, an abdominal and transfontanelle ultrasounds were performed. The transfontanelle examination (Figures 20 and 21) observed morphoestrutural abnormalities of the supra-tentorial ventricular system, not characterizing the third ventricle and visualized the presence of echogenic component in bilateral occipito-parietal posterior cortical projection being indicated head CT scan or MRI. On abdominal examination, there was hepatomegaly, splenomegaly, polycystic kidneys with bilateral hydronephrosis (Figures 18 and 19) and severe ascites.

In two weeks, a head CT scan (Figures 22 and 23) was performed, which visualized convergent strabismus, ventricular systems dilatation, not visualizing the septum pellucidum - alobar holoprosencephaly, enlarged subarachnoid space and absence of cortical gyri - lissencephaly, overlapping skull bones and hyperdense material bordering the cerebellar hemispheres, questioning blood.

Karyotype test was done by G banding, confirming the trisomy of 13 chromosome with the karyotype 47, XX, +13.
Case 2

Male patient, 05 months old, brown, brazilian, born and raised in Santos (SP), being admitted to the pediatric ICU of Santa Casa da Misericórdia de Santos, acute respiratory failure (was already in Home Care). On physical examination presents anicteric, afebrile, pale, dehydrated, hypoactive and spastic. Cardiac auscultation sounds were rhythmic in 2 times with systolic murmur 2 + / 4 +, with a heart rate of 116 bpm. Auscultation with rhonchi and crackels with respiratory rate of 38, saturating around 93%. Blood pressure of 100 X 70 mmHg. Fontanelle flat and normotensive. Abdomen was distended, with a palpable liver 3 cm from the right costal margin. Note, also on physical examination, some peculiarities such as facial hirsutism, micrognathia, triangular head, bilateral inguinal and umbilical reducible hernia, overlapping hallux and especially clenched fists with overlapping fingers of both hands (Figure 24).

Born by cesarean section having 40 weeks and 5 days by last menstrual period, with Apgar score at 1 minute of 2, 5 minutes of 6 and in ten minutes of 9, weighing about 2580 grams, 35 cm head circumference and 43,5 cm in length. Capurro compatible with 37 weeks and 1 day. Moved to ICU due to acute respiratory failure, was intubated and also where, after 05 days of hospitalization, had seizures - so began the use of phenobarbital. Was hospitalized for about 03 months. However, soon after discharge, he was again admitted to the ICU due to cardio-respiratory arrest, being hospitalized for 10 days.

Mother, 39 years old, had gestational hypertension controlled with methyldopa, nurse, denies smoking and drinking, having previous uneventful pregnancy and a abortion. Serum creatinine clearance of 0.7 mg/dl and 24-hour proteinuria of 66 mg/24hrs. 7 held consultations in prenatal, showing no complications. Serology for HIV, IgM toxoplasmosis, Hepatitis C and IgM rubella negative. IgG toxoplasmosis, anti-HBs and IgG rubella were reagents. Father with 33 years old, hypertensive and overweight, pharmacist, social drinker, denies smoking. There is no consanguinity between the parents and grandparents, both paternal as maternal. Paternal grandfather was diabetic.

During pregnancy, the first two ultrasound exams were normal. However, in the third test, about 29 weeks according to last menstrual period was observed intrauterine growth retardation, because it had, under the action of the test, about 26 weeks and 05 days. Four weeks later, in addition to growth restriction, added weight is below the 10th percentile for gestational age. After another four weeks, the weight was below the 5th percentile for gestational age, and missing two weeks for birth, also presented pyelocalyceal system discrete dilatation in the right kidney (Figure 25).

About 21 weeks of gestation, according to date of last menstrual period, the morphologic ultrasound (Figures 26-38) showed the stomach filling mild, even with reassessment
after 55 minutes and slightly enlarged gallbladder, being esophageal atresia questioned - unconfirmed after birth. Rest of the exam was normal, including the umbilical cord that had three vessels (two arteries and one vein). Also, it was indicated genetic counseling and fetal karyotype research.

Two-Dimensional Color Doppler Echocardiography (Figures 39-43) performed at birth in which visualized atrial septal aneurysm with ostium secundum atrial septal defect measuring approximately 0.16 cm, with directed flow from the left to the right atrium and sinus venosus atrial septal defect measuring approximately 0.23 cm with directed flow from the left to the right atrium. Patent ductus arteriosus with flow directed from pulmonary artery to the aorta. It is also envisioned, transposition of the great arteries. Interventricular communication inlet type measuring 11.5 mm with flow from the left ventricle to the right, with mild dilatation of the right cardiac chambers.

The patient progressed with infection (neonatal sepsis), being treated with antibiotics - ampicillin, amikacin and cefotaxime. During hospitalization, due to the history of seizures, a polysomnography was performed which showed slowing and disorganization of brain electrical activity-based, with the presence of unusual activity, polymorphic, often exacerbated organized in brief spurts of 2-3 seconds length in the central regions-bilateral midline projection.

With 22 days of life, he underwent a cardiac surgery with pulmonary artery bandage and ductus arteriosus ligation, being extubated after 09 days. However, after surgery, the patient presented again with sepsis being treated with antibiotics in the ICU. When starts pediatric monitorization, had difficulty breastfeeding orally, depending of enteral feeding, difficulty in weight gain and hemodynamic controlled by medications. Discharged to Home Care, with 103 days of life.

In evolutionary control of Two-Dimensional Color Doppler Echocardiogram with 04 weeks of postoperative pulmonary banding and closure of the ductus arteriosus was noted decrease of interatrial and interventricular communication, ductus arteriosus occlusion and pulmonary artery banding (Figures 44-47).

Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery, which showed large ventricular septal defect, perimembranous, subaortic extending to the inlet and trabecular muscle with left-right flow through the orifice (Figures 48-52). Also showed slight concentric hypertrophy of the left and right ventricles, besides the banding in the middle of the pulmonary trunk.
Head CT scan performed with 05 months of life (Figures 53 and 54) indicates dilatation of the ventricular system, with an area of hypoattenuation coefficient similar to liquor in bilateral external capsule projection and an accentuated increase of ventricular spaces.

The abdominal ultrasound with 06 months of life (Figures 55 and 56), found a parenchymatous nephropathy, with bilaterally mild hydronephrosis with an empty urinary bladder. Urea: 42 mg/dl. Creatinine: 0,23mg / dl.

After birth, karyotype test was done by G banding, confirming the trisomy of 18 chromosome with the karyotype 47, XY, +18.

**COMPARISON OF THE PATIENTS FINDINGS**

**CLINICAL FINDINGS**

<table>
<thead>
<tr>
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<tr>
<td>Bradycardia</td>
<td>Pale and dehydrated</td>
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<tr>
<td>Microcephaly</td>
<td>Micrognathia and triangular head</td>
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<tr>
<td>Absence of nose</td>
<td>Hypoactive and spasticity</td>
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<tr>
<td>Extense cleft palate</td>
<td>Hirsutism on face</td>
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<tr>
<td>Ambiguous genitalia</td>
<td>Bilateral inguinal hernia and umbilical hernia</td>
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<td>Umbilical stump with one artery and one vein</td>
<td>Seizures</td>
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<tr>
<td>Polydactyly in both hands</td>
<td>Clenched fists with overlapping fingers of both hands</td>
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**SONOGRAPHIC FINDINGS**

<table>
<thead>
<tr>
<th>PATAU</th>
<th>EDWARDS</th>
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<tbody>
<tr>
<td>Intrauterine growth retardation</td>
<td>Intrauterine growth retardation</td>
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<tr>
<td>Umbilical cord with single umbilical artery</td>
<td>Stomach filling mild, even with reassessment after 55 minutes, being esophageal atresia questioned</td>
</tr>
<tr>
<td>Holoprosencephaly</td>
<td>Slightly enlarged gallbladder</td>
</tr>
<tr>
<td>Microcephaly</td>
<td>Pyelocalyceal system dilatation in the right kidney</td>
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</tbody>
</table>
Bilateral cleft lip
Polycystic kidneys
Absence of nose
Hypotelorism
Probable esophageal atresia.
Polydactyly in both hands

**ECHOCARDIOGRAPHIC FINDINGS**

**PATAU**
- Aorta riding the ventricular septum
- Right ventricle with reduced dimensions
- Enlarged left ventricle
- Communication between the ventricular

**EDWARDS**
- Transposition of the great vessels
- Dilatation of the right heart
- Communication between the atria
- Communication between the ventricular

**CT SCANS FINDINGS**

**PATAU**
- Increased subarachnoid space and absence of cortical gyri - lissencephaly
- Ventricular system dilatation
- Hyperdense material bordering the cerebellar hemispheres, wondering blood.
- Alobar holoprosencephaly
- Overlapping skull bones
- Convergent strabismus

**EDWARDS**
- Large increase in ventricular spaces.
- Ventricular system dilatation
- Area of hypoattenuation coefficient similar to liquor in bilateral external capsule projection
Fig. 1: Umbilical cord with single umbilical artery.

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Fig. 2

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Fig. 3: Reverse wave A of the ductus venous Doppler.

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Fig. 5: Polydactyly.

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Fig. 12: Hypotelorism.

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Fig. 11: Polydactyly.

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Fig. 10: Cleft lip.

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Fig. 9: Nose agenesis.

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Fig. 8

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Fig. 7: Holoprosencephaly.

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Fig. 6: Single umbilical artery.

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Fig. 13: Microcephaly.

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Fig. 14: Fetal echocardiographic doppler.

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**Fig. 15:** Fetal echocardiographic doppler.

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Fig. 16: Fetal echocardiographic doppler.

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Fig. 17: Fetal echocardiographic doppler.

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Fig. 20: Transfontanelle ultrasound.

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Fig. 18: Polycystic kidneys with bilateral hydronephrosis

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Fig. 19: Polycystic kidneys with bilateral hydrenephrosis.

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Fig. 21: Transfontanelle ultrasound.

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Fig. 22: Head CT Scan.

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Fig. 23: Head CT Scan.

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Fig. 37: 21 weeks of gestation morphologic ultrasound.

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**Fig. 36:** 21 weeks of gestation morphologic ultrasound.

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**Fig. 35:** 21 weeks of gestation morphologic ultrasound.

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Fig. 34: 21 weeks of gestation morphologic ultrasound.

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Fig. 33: 21 weeks of gestation morphologic ultrasound.

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Fig. 32: 21 weeks of gestation morphologic ultrasound.

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**Fig. 31:** 21 weeks of gestation morphologic ultrasound.

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Fig. 30: 21 weeks of gestation morphologic ultrasound.

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Fig. 29: 21 weeks of gestation morphologic ultrasound.

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**Fig. 28:** 21 weeks of gestation morphologic ultrasound.

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**Fig. 27:** 21 weeks of gestation morphologic ultrasound.

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**Fig. 26:** 21 weeks of gestation morphologic ultrasound.

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**Fig. 24:** Clenched fists with overlapping fingers of both hands.
Fig. 25: Pyelocalyceal system discrete dilatation in the right kidney

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Fig. 38: 21 weeks of gestation morphologic ultrasound.

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Fig. 39: Two-Dimensional Color Doppler Echocardiography performed at birth.

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Fig. 40: Two-Dimensional Color Doppler Echocardiography performed at birth.

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**Fig. 41:** Two-Dimensional Color Doppler Echocardiography performed at birth.

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**Fig. 42:** Two-Dimensional Color Doppler Echocardiography performed at birth.

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Fig. 43: Two-Dimensional Color Doppler Echocardiography performed at birth.

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**Fig. 44:** Two-Dimensional Color Doppler Echocardiogram with 04 weeks of postoperative pulmonary banding and closure of the ductus arteriosus.

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Fig. 45: Two-Dimensional Color Doppler Echocardiogram with 04 weeks of postoperative pulmonary banding and closure of the ductus arteriosus.

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Fig. 46: Two-Dimensional Color Doppler Echocardiogram with 04 weeks of postoperative pulmonary banding and closure of the ductus arteriosus.

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Fig. 47: Two-Dimensional Color Doppler Echocardiogram with 04 weeks of postoperative pulmonary banding and closure of the ductus arteriosus.

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Fig. 52: Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery.

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**Fig. 51:** Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery.

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**Fig. 48:** Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery.

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**Fig. 49:** Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery.

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**Fig. 50:** Two-Dimensional Color Doppler echocardiogram performed 5 months after surgery.

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Fig. 53: Head CT scan performed with 05 months of life.

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Fig. 54: Head CT scan performed with 05 months of life.

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Fig. 55: Abdominal ultrasound with 06 months of life.

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Fig. 56: Abdominal ultrasound with 06 months of life.

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Conclusion

The definitive diagnosis of both syndromes is still karyotype. However, the diagnosis of each syndrome varies between sonographic findings, with holoprosencephaly being more favorable of Patau Syndrome and clenched fists with overlapping fingers for Edwards Syndrome.
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