Abstract

Patau syndrome was first described in 1960 as a group of birth defects caused by trisomy of chromosome 13 (T13). Providing accurate information and relevant reproductive genetic counseling that would allow parents to make informed decisions is not easily accomplished because of the limited information available prenatally. Only 1/3 of all cases of T13 are diagnosed prenatally, which means it cannot be expected that most cases will be detected early in pregnancy, that the parents will decide to terminate the pregnancy, and that difficulties will be avoided. There is no good prenatal screening for T13, and there are many kinds and degrees of anomalies. About 60% of cases are first detected in the second trimester, and life expectancy is difficult to predict. When patients choose not to terminate pregnancy, or when the pregnancy has progressed to a viable gestational age, pregnancy termination is no longer an option. Also, nowadays 12% of couples choose to continue pregnancy following chromosomal confirmation of a suspected T13. The aim of this work is to elucidate for health care providers what problems they are likely to face in the care of children with T13 and in contact with their parents. It is crucial for the management of each case to discuss neonatal procedures of resuscitation, alternatives to aggressive resuscitation, the possibilities for correcting some of the defects, and to be prepared to guide the parents through the trauma of having a child with a lethal defect (Adv Clin Exp Med 2015, 24, 5, 911–921).

Key words: Patau syndrome, trisomy 13, holoprosencephaly, aneuploidy.
a high mortality rate. Newborns with mosaic variations tend to be less severely affected [10]. Out of 27 cases of Patau syndrome investigated by Taylor [11], 70.4% had primary trisomy 13-15 (D), 11.1% had D/D interchange trisomy, 3.7% were mosaic, 3.7% had 46 chromosomes with a deleted short arm of a B chromosome [46, XY, Bp-] and 7.4% had normal chromosomes [11]. Triploidy is associated with early pregnancy loss; it rarely lasts beyond 20 weeks [12]. Trisomy 13 is due to chromosomes that did not disperse during meiosis in one of the parents. As Pilu and Nicolaides noted, “When there is a double maternal chromosome contribution, the pregnancy may persist into the third trimester” [12]. According to various sources, trisomy 13 occurs with a frequency between 1 in 5000 and 1 in 14,500 live-born children [13–16]. Newborns with trisomy 13 have many abnormalities, involving nearly every organ, as well as developmental delay.

Because T13 occurs so rarely, only collected data reveal the spectrum of clinical symptoms of the syndrome [11]. Taylor’s survey also showed which features occur both in Patau and in Edwards’ syndrome (trisomy 18), and which appear only in one or the other [11]. The gender distribution in Edwards’ syndrome is grossly uneven, with females prevailing, but in Patau syndrome it is much closer to normal. The mean survival time is similar for both sexes, although longer in females [11]. Maternal age is relevant in both syndromes (Fig. 2): The risk of trisomies increases with maternal age [10–12]. For example, when the mother is 20 years old, the risk of trisomy 13 in week 10 of gestation is 1/6347 and in week 40 it is 1/42,423. When the mother is 35 years old, the risks for the same weeks of gestation are, respectively, 1/1481 and 1/9876; when she is 42 years old, the risk rises significantly, to 1/227 in week 10 and 1/1516 in week 40 [17]. This means that the risk of trisomy 13 increases with maternal age, but it drops with gestational age, as fewer fetuses with trisomy 13 survive until late gestational age.

The newborn’s prognosis is bad mainly because of central nervous system (CNS) and cardiac defects [2]. The few infants who survive the first six months of life usually die due to heart failure or concomitant infections. Most affected children die before the age of one year; fewer than 10% survive the first year [11, 18, 19]. Statistically, in 74 cases of Patau syndrome, the mean survival time was 89.2 ± 29.9 days [11]. There have only been a few cases in which the children survived to 10 years [20]. Trisomy 13 and 18 are associated with a high risk of miscarriage or stillbirth – about 80% [12, 21], while about 15% of all pregnancies end in spontaneous abortion [22]. A case has also been reported of a neonate born after term (week 43) who weighed 3240 g [6]. The risk of fetal loss (i.e., spontaneous abortion or stillbirth) following prenatal diagnosis of trisomy 13 for the period between the 12th and 40th weeks of gestation is 49%;

Fig. 1. Patau syndrome – cleft lip and palate karyotype: simple trisomy 13 [43]

Fig. 2. Maternal age distribution in (a) Edwards’ syndrome, (b) Patau syndrome, (c) Down syndrome, and (d) the general population [11]
for the period between the 18th and 40th weeks it is 42%; and for the period between the 24th and 40th weeks it is 35% [23]. This risk is greater for male fetuses [23].

Of the three autosomal trisomy syndromes – Down’s syndrome (trisomy 21), Edwards’ syndrome (trisomy 18), and Patau syndrome (trisomy 13) – the last two are rare and many of the characteristic features occur in both of them. After calculating the discriminating ratio Taylor used two arbitrary cut-off points to identify three areas: “Patau features” (< 0.5), an area of major overlap (0.5 to 1.5), and “Edwards’ features” (> 1.5) [11]. Patau’s features include microcephaly and eye defects (microphthalmos, iris colobomata), low-set ears and facial defects (hare-lip, cleft palate), foot and hand defects (polydactyly, long hyperconvex finger-nails, talipes equinovarus, fibular S-shaped hallucal arch) and capillary hemangiomma [11]. The area of major overlap (0.5–1.5) includes ocular hypertelorism, presumptive deafness, epicanthic folds, strabismus, micrognathia, low-set malformed ears, short neck, extra skin on the nape, flexion deformity of the fingers, single palmar crease, inguinal/umbilical hernia, congenital heart disease, cryptorchidism in males, bicornate uterus in females, feeding difficulty, jaundice, developmental retardation, failure to thrive, hypotonia, jitteriness, apnea and seizures [11]. Abnormal fingertip patterns and positions of the palmar triradii are also noted. Sometimes the fingers are abnormally flexed. A distally placed triradius and fibular S-shaped hallucal arch have occurred [11]. In both these syndromes, infants are characterized by low body weight and low height, and are often born prematurely.

Trisomy 13 is also correlated with placental abnormalities, such as preeclampsia (PE), molar placenta, reduced placental vascularization, small placental volume, placental mesenchymal dysplasia (the PE-causing gene on chromosome 13) [5, 12, 24–26].

Nowadays the use of diagnostic schemes based on ultrasound, biochemical and cytogenetic tests increases the likelihood of diagnosing chromosomal aberrations during pregnancy, even in early pregnancy [12, 13, 27–29]. However, most parents do not realize that, unlike the situation with Down syndrome, methods for prenatal diagnosis of trisomy 13 and 18 are still insufficient. In a study by Parker et al., out of 44 T13 cases and 88 T18 cases, 64% were first detected through chromosomal analysis initiated because of abnormalities noted on fetal anomaly scanning in the second trimester, whereas only 3% of cases were detected through the serum-screening program currently offered for Down syndrome. In 11% of cases the diagnosis was first suspected after birth [10]. In a sample of 24 cases of trisomy 13, only one third were diagnosed prenatally and only one was electively terminated [9]. To change these proportions, a highly sensitive prenatal screening program is needed; however, due to parents’ individual decisions, this is not tantamount to saying that the number of pregnancy terminations will increase significantly.

Later in pregnancy, in the 18–23-week scan, trisomy 13 demonstrates major and minor defects: holoprosencephaly (HPE) and associated facial abnormalities (facial cleft), microcephaly, posterior fossa cyst, enlarged cisterna magna [12]. Therefore, while performing USGs, it should be borne in mind that prenatally detected malformations of the CNS are strongly correlated with chromosomal abnormalities. Different isolated malformations indicate different syndromes: Isolated HPE, spina bifida and agenesis of corpus callosum are significantly associated with trisomy 13; isolated anencephaly is associated with trisomy 18; and the only CNS abnormalities associated with trisomy 21 are ventriculomegaly and choroid plexus cyst [30]. Also among the major and minor defects in trisomy 13 are cardiac and renal abnormalities (often enlarged and echogenic kidneys, mild hydronephrosis), nuchal edema, diaphragmatic hernia, exomphalos and postaxial polydactyly, and talipes [12].

HPE is a congenital defect of the forebrain, median structures and face resulting from incomplete cleavage of the brain during early embryogenesis. The most severe variant of HPE is alobar HPE [31]. In humans, HPE occurs in patients with trisomy 13, trisomy 18, trisomy 13-15, trisomy 13-15 mosaicism, ring chromosome 13 or 18, and chromosomal deletion 13 or 18, in Klinefelter syndrome and trisomy 10, but maternal diabetes mellitus and fetal alcohol exposure should always be excluded [2, 32–34]. Among newborns with trisomy 13, 24–45% have HPE, and trisomy 13 accounts for 60–75% of cases of patients with HPE due to chromosomal anomalies (including cryptic rearrangements) [2, 32, 35]. Other authors estimate that 17–39% of patients with trisomy 13 have HPE [36], these lower estimates are the result of prenatal diagnosis by USG. Pathologic examination, which is the most objective, shows that as many as 67% of patients with trisomy 13 have signs of forebrain anomalies consistent with HPE [37]. The most common clinical features of trisomy 13 include mental and growth retardation, craniofacial dysmorphisms, hand and foot anomalies, brain, heart and kidney defects, which are also found in carriers of 13q partial deletion [32].

Trisomy 18 is much less often seen in conjunction with HPE (1–2%) [32]. The presence of ZIC2 on chromosome 13, another locus within
13q22-33 (different from ZIC2), and the association of this gene with HPE and HPE-spectrum anomalies are still under examination [38]. Four genes related to brain development and their chromosomal regions have been studied: HPE1, HPE2, HPE3 and HPE4, and the broad spectrum of HPE phenotypes reflects the complexity and heterogeneity of its etiology [39].

These observations require further research because, as Ballarati et al. wrote, “patients carrying deletions of precisely the same chromosomal regions may show different phenotypic features depending on the genetic background in which they are placed” [38]; for example, the incidence of HPE is about 0.48–0.88 per 10,000 live births with normal chromosomes [32, 40]. There are three types of HPE: alobar, semilobar and lobar [41], with alobar being the most severe variant (Fig. 3) [2, 42]. Kidney defects are noted in about 30% of children with trisomy 13, omphalocele in 10%, and skin defects of the scalp in 20% [35]. About 80% of infants with Patau syndrome have a congenital heart disease (ventricular septal defect, atrial septal defect, patent ductus arteriosus, dextrocardia, hypoplastic left heart syndrome). Medical problems after birth depend on the type of congenital malformation. For example, difficulties with breathing are due either to apnoe or problems with lung development; problems with feeding arise when there is malformation of the palate, while gastroesophageal reflux predisposes to aspiration and, as a consequence, to aspirational pneumonia [2, 35]. Usually the newborn presents respiratory distress syndrome (RDS) requiring high levels of oxygen supplementation, progressing to bradycardia and apnea [2]. In infants and young children, other common disorders are motor disability, developmental disability, hearing loss, slow postnatal growth, seizures and hypertension [35]. Malformations of the eyes (anophtalmia, microphthalmia, hypotelorism or cyclops) (Fig. 4) and polydactyly (Fig. 5, 6) [43] are so common that they might be called hallmarks of the syndrome. Sometimes faces look bizarre (e.g., like a trumpet, Fig. 7) [43].

It is difficult to predict the life expectancy of a baby with trisomy 13 if there are no immediate life-threatening problems. The more severe the defect, the sooner fetal loss occurs, due to miscarriage, stillbirth, spontaneous abortion or very premature labor with early death after birth.

The risk of trisomies in women who previously had a fetus or child affected by trisomy is higher than in the general population [17]. For example, in a 35-year-old woman who had a baby with trisomy in a previous pregnancy, the risk at 12 weeks of gestation increases from 1/249 (0.4%) to 1/87
Also, a personal or close family history of giving birth to an affected child increases the risk of trisomy 13 [10]. The possible mechanism for this increase is the fact that about 5% of couples who had an affected child in a previous pregnancy or in their close family history “have parental mosaicism or a genetic defect that interferes with the normal process of disjunction but in 95% the risk of recurrence is not actually increased” [17].

However, for the parents the only thing that matters is that the child will have defects, that they do not know how long it will survive or whether they can cope with it. These psychological problems are vividly depicted in the following passage: “Parents who discover during pregnancy that their baby has a serious chromosomal condition such as Patau syndrome suddenly find themselves on an unexpected journey away from their hopes for a healthy baby and a normal family life, towards a new and challenging situation” [18].

Progress in fetal medicine allows for early diagnosis and makes it possible to terminate pregnancy, which is allowed by Polish law. However, the parents’ decision about whether to terminate or to continue the pregnancy is influenced to a high degree by the way the situation is presented to them by the doctor. It is important to remember that “nondirective counseling before prenatal diagnostic testing does not require a patient to commit to pregnancy termination if the results are abnormal” [44].

Parents should also be advised that if the child is born alive, there are various ways in which some anomalies can be corrected. Cardiac surgeons, maxillofacial surgeons or plastic surgeons can do a lot. With primary or secondary cleft palate, which frequently occurs in children with Patau syndrome, orthodontic treatment is needed in order to facilitate feeding. For example, there have been cases in Poland when such orthodontic treatment was initiated in four- and six-day-old patients with a palatal plate due to primary and secondary bilateral cleft palate [45].

The survival of an infant with Patau syndrome depends mainly on the degree of CNS anomaly and heart defects. The median age at death for infants with trisomy 13 is seven days, and only 5.6% of children the syndrome survive to the age of one year or older [46]. Median age at death is higher for girls than for boys, and higher for African American children than for non-African American children [46]. However, even when heart defects are corrected surgically, the survival age is not significantly increased [47]. Cases of several years’ survival are very rare, but they do happen; there have been dozens of such individuals in the world. Some of those children are still alive, and are trying to catch up with their healthy peers. Jenna Cole (Fig. 8) belongs to this small group; she practices artistic gymnastics and achieves very good results [48].
Pregnancy management in cases of fetuses with trisomy 13 should take place in hospitals that are prepared to it, not only as far as the gynecologists’ competence is concerned, but also in terms of offering appropriate psychological counseling to the parents. When the parents choose to continue the pregnancy, or when they postpone the decision until the gestational age is reached when abortion is no longer an option, the obstetrician’s attention should concentrate on choosing the time and method of delivery [49]. In cases of lethal conditions, “tocolysis in an effort to prevent preterm birth is not a reasonable option” [49]. The decision about the method of delivery is determined by obstetrical considerations, as in all other pregnancies, and does not depend on the presence of the syndrome. It should be remembered, however, that “cesarean delivery for fetal indications is not recommended due to the lethal nature of this condition” [49]. The overall plan of management should include a discussion with neonatologists on “neonatal procedures of resuscitation, the cost of these measures, and alternatives to aggressive resuscitation” [49]. Opinions on the viability of aggressive resuscitation are divided, even among the authors of this article.

The recommendation that the hospital should be prepared to care for children with trisomy 13 becomes reality only when the medical staff is confronted with the specific child and his or her parents. First, the parents, who have heard of great advances in medical care and prenatal screening, are disappointed that Patau syndrome is recognized late in the pregnancy, after the legal time for abortion has passed. What they do not know is that popular education and discussions are practically always limited to Down syndrome. Diagnosing Patau syndrome not only results in the parents’ disappointment, but also puts them and the health service units in a special situation. Time is needed for the parents to accept their new situation, and simultaneously the health care units must be prepared to act quickly, meeting the highest standards of professional care.

How difficult this can be was demonstrated by a case in which the authors of the present article were involved. The case was the first pregnancy of a 24-year-old woman, an economist in good health and with good social and living conditions. She did not use alcohol or drugs and was not a smoker. She was negative for syphilis, HIV and hepatitis B. The father was 29 years old, also an economist with no bad habits and in good health. There had been no children with genetic defects in the families of either of them.

The risk of trisomy 13 rises as maternal age increases, but not as markedly as with Down syndrome or Edwards’ syndrome [10]. In cases where the mother is young, both the patient and the doctors may relax their vigilance. Apart from that, medical history is essential. Several maternal and environmental factors (for example diabetes mellitus or alcohol) are associated with CNS abnormalities that usually constitute the basis for genetic diagnostics [40]. In experimental studies, both alcohol and the ingestion of certain plants induced facial and CNS abnormalities in animals [40].

Even nowadays, when prenatal diagnostics has made incredible progress, there are cases in which children are born with malformations despite the implementation of a number of tests revealing no fetal malformations. In the case dealt with by the authors of this article, the patient had proper prenatal care (more than ten medical examinations during the pregnancy, six ultrasound scans and two 4D ultrasound scans). On the basis of the experience of the authors of this article, for the majority of pregnant women in Poland having a 4D scan is more important as a way to obtain a souvenir for the family album than as a way to insure a proper diagnosis. It can only be regretted that in the case described here morphological malformations were not recognized (Fig. 9).

The first bad news raising suspicion of some kind of chromosomal anomaly was found in the 33rd week, when oligohydramnion and intrauterine growth retardation were diagnosed. At the gestational age of 36 weeks and 4 days, a female fetus was delivered by cesarean section, due to fetal distress syndrome. She weighed 1830 g and was 50 cm long; her Apgar score was 8/9/9 at 1/5/10 min. There were visible symptoms of genetic disease: omphalocele,
a cleft soft palate, loss of skin on the top of the head, microphthalmia, extra fingers and toes, hypotrophy and low-set ears (Fig. 10–14). A probe was introduced into the stomach. After several hours, the infant was transferred to the Department of Pediatric Surgery, where plastic surgery of the front abdominal wall was performed, the navel was reconstructed and extra fingers were amputated.
The newborn presented RDS and was referred to the pediatric emergency care unit. During the hospitalization there were difficulties with ventilation. For a few days high frequency oscillatory ventilation (HFOV) was conducted and improvement was achieved. The patient underwent an extubation attempt, which after a few hours turned out to be unsuccessful. On day 11, the baby was in serious condition but stable, breathing inefficiently and intubated, and was referred to the ICU for further treatment. The child was placed in an incubator, breathing with a Servo ventilator. Material was collected for cytogenetic blood testing. Infants with Patau syndrome and Edwards’ syndrome can have similar features, and it is difficult to differentiate between them after birth. In this case, unbalanced Robertsonian translocation was found, so trisomy 13 was diagnosed. Genetic counseling and genetic testing were offered to the family.

Robertsonian translocations (RT), also called whole-arm translocations or centric-fusion translocations, are “a rare form of chromosomal arrangement that in humans occurs in the five acrocentric chromosome pairs: 13, 14, 15, 21 and 22” [50]. About one in a thousand newborns has Robertsonian translocation [51]. “A Robertsonian translocation in balanced form results in no excess or deficit of genetic material and causes no health difficulties. In unbalanced forms, Robertsonian translocations cause chromosomal deletions or additions and result in syndromes of multiple malformations, including trisomy 13... and trisomy 21” [50].

A child with a congenital defect can cause a serious family crisis and disruption of a normal parental relationship with the child. The first task for the parents is to reconcile the idealized image of the child they expected with the reality. Acceptance of the child by the parents and their adaptation to the new situation depends not only on the personality of each of them, their attitudes, social background and prior experience, but above all on the relationship between them, and on their general ability to cope with difficult situations. The reaction of the parents to the birth of a child with defects also depends on the type of defects. The reactions of mothers who give birth to children with visible facial defects, such as cleft lips, are stronger than those who have babies with cleft palates [18]. The intensity of the reaction also depends on whether the defect can be removed, whether it is incurable or life-threatening. It also depends on whether the child has one defect or multiple defects, and whether other family members have similar defects. Parents mourn the loss of an imagined, often long-awaited, child before establishing an emotional relationship with the one they really have. If the child is alive, his or her parents constantly face problems, such as the child’s mental retardation. Newborns with CNS anomalies who survive usually have mental retardation and often present epilepsy [2]. If the CNS anomaly involves the hypothalamus, the child may have diabetes insipidus, and/or poikilothermy may be present [2].

The reactions of the parents are characterized by grief and chronic stress. The stress means that some information about the child’s health does not get through to them. Usually the parents of a child with a congenital condition go through specific phases of complex emotional reactions, but their intensity and duration vary. The first phase is a period of shock. Some parents do not respond with shock, but try to rationalize the problem and focus on the facts related to their child’s illness. The next phase is the period of disbelief, when most parents show negative reactions of varying intensity. After this phase, the phase of grief and anger occurs. The parents are angry with the doctors, saying they were sure the prenatal care was of a high standard but are now convinced it was not so. One of the things they usually do not realize is that even if the patient had been sent for genetic amniocentesis after the 24th-week scan and trisomy 13 had been diagnosed, the risk of miscarriage or stillbirth between the 24th and 40th week is 35% in cases of trisomy 13 [23], so the pregnancy would not have been terminated, but their stress would have begun earlier. Nothing justifies an assumption that the stress could have been lessened. The parents would have been informed that, for example, first-year survival rate calculated in Texas is the highest for infants with gastroschisis (92.9%), trisomy 21 (92.3%), cleft lip with or without cleft palate (87.6%), but almost all infants with trisomy 13 or
Trisomy 18 die during the first year of life [52]. Facial cleft without other abnormalities increases the odds of dying during the first year of life 3.7 times, but when it is associated with other abnormalities, the odds increase 82.3 times [53].

In the authors’ experience, it is more difficult to inform parents about trisomy 13 after the child is born, when the parents have already seen the child, their parental feelings have developed and they hope for the child’s survival. Is there a good way to tell them that the highest risk of death is during the neonatal period (77% deaths occur during the first 27 days), and overall first-year survival for infants with congenital anomalies is low [52]. On the other hand, if there is no serious CNS or heart defect, a child can live more than ten years, as Jenna has [48]. These estimates can be useful in counseling women who are carrying an affected fetus, but they are not at all useful for guiding parents through the short period of their parenthood. The aim of the article is not only to help doctors make decisions, but also to voice support for prenatal screening programs. At the same time in should be noted that genetic examinations in the first trimester do not always provide a complete diagnosis [17]. There was a case of non-mosaic trisomy 13 [46, XX, der(13;13)(q10;q10)] where subsequent amniocentesis and cordocentesis showed varying percentages of abnormal cells and mosaic trisomy 13. After the birth, karyotyping of chorion cells showed indirect evidence of trisomy 13 coexisting with euploidy [46, XX], aneuploidy [16, XX-13, +mar], and monosomy 13 [45, XX, -13]. The examination of the newborn showed low-set ears, no 12th rib, cardiomegaly with a septal defect and abnormal karyotypes of skin fibroblasts. Blood lymphocytes and cardiac tissue were normal [46, XX]. The baby survived beyond 8 months [47].

The authors are looking forward to widespread application of maternal plasma cell-free DNA sequencing [54]. This tool is non-invasive for fetuses in high-risk pregnancies, and detects nearly all cases of Down syndrome, trisomy 18 and trisomy 13. The false-positive rates are 0.28% for trisomy 18 and 0.97% for trisomy 13 [54].

Coming back to difficult decisions and the parents’ situation, in the authors’ experience one can never be prepared for the news that one’s newborn child has a severe genetic defect such as Patau syndrome. Sometimes women tear up ultrasound images and throw out keepsakes of the baby, because immediately following the news they want everything related to the baby out of sight in order to reduce their pain. Later they achieve relative emotional balance and often look for the discarded keepsakes. The husband also goes through the equally tragic experience of suddenly being deprived of fatherhood. Even though he feels emotional pain, he tries to support his wife with all his strength. In some Western European countries, in such situations midwives take the infant’s footprints and handprints, and cut off little snips of hair and put them in a folder among the documents. If the mother later expresses a wish to take these keepsakes home – which is not always the case – she can do so. In Poland there is no such practice. At first, the parents’ shock is huge. It is hard to clearly identify the emotional state that the parents experience after learning that their child has Patau syndrome. Bereavement is generally associated with the state after the death of a living person. Grief is rarely refers to the loss of an unborn child, and even more rarely when a malformed child is born. When coping with difficult situations, it seems that most parents have one thing in common: their inability to process and absorb all the information that they demand when they become aware of their child’s condition. They ask questions and receive answers, but the answers are not heard, or rather not understood. They receive information, but their mind does not always process it. It is difficult for them to accept the diagnosis. A lot depends on how the information is presented to them. Appropriate behavior from the members of the hospital team is crucial here, as is emotional support for the parents in difficult situations [18].

Another issue is offering the parents appropriate counseling about the risks involved in the next pregnancy. As Best put it: “All patients diagnosed prenatally with a fetus affected by Patau syndrome should be offered a consultation with a care provider skilled in delivering serious information who is knowledgeable about recurrence risk, screening, and diagnostic testing options for future pregnancies.... [A] genetics counselor is an ideal source (…)” Once a diagnosis of Patau syndrome is made, pregnancy management varies according to the gestational age at diagnosis. At preivable gestational ages, the option of pregnancy termination should be among those discussed” [49]. The gestational age limits for termination vary from country to country. However, the matter should not be left to the obstetrician alone, as some of them may follow the path that seems the easiest from a purely “technical” point of view: It is easiest to terminate pregnancy, and recommending that solution may seem the simplest. There are doctors who go even further, carrying out the termination of pregnancy through hysterectomy in a patient at the age of 42 [55]. This is why most authors agree that the obstetrical and genetic aspects of trisomy 13 should be expanded to include ethical considerations, and parents at each stage should be provided with appropriate psychological care.
References


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