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Original Article

Prenatal Ultrasound Diagnosis and Short-term Outcome of Congenital Malformations: Experience of the Maternity and Reproductive Health Hospital "Les Orangers" - Rabat, Morocco, between 2011-2016

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ABSTRACT

Background: The objective of our study to analyze the data of the prenatal diagnosis of congenital malformations at the maternity and reproductive health hospital "Les Orangers" in Rabat and to identify the main anomalies detected, their percentage and their short-term outcome.

Methods: This is a cross-sectional study conducted at the maternity and reproductive health hospital "Les Orangers" in Rabat, Morocco. The data was collected and reported on pre-established sheets and on the register of malformations of the hospital.

Results: A total of 245 cases of congenital malformations comprising 470 types of congenital malformations were recorded out of a total of 43,923 births over a period of five and a half years, giving a prevalence of 5.58 per thousand.

Prenatal diagnosis was made in a third of cases (33%), essentially during the 2nd-3rd trimester of pregnancy. The anomalies revealed by this antenatal diagnosis were dominated by urinary malformations in 70% and central nervous system anomalies in 67%, followed by other types of congenital anomalies in less than 40% of cases, while genetic problems were detected in 2.5%; this rate is underestimated since chromosomal abnormalities sometimes appear as syndromes, so that in some diseases, genetic changes are not separated from other abnormalities, and since more than half (50.7%) of cases presenting polymalformative syndromes not survived 77% of cases i.e. (48.5% of deaths and 28.5% of FDIU) and an etiological study was not carried out.

Conclusion: Antenatal ultrasound allows early detection and monitoring of the evolution of congenital malformations, and thus the possibility of ensuring early and adequate management of these anomalies from birth. In our context, it is necessary to develop a prenatal screening program for congenital anomalies and a network of reference centers for the management of these anomalies in order to improve their prognosis.

Keywords: Antenatal diagnosis, Congenital malformation, Evolution, Morocco

Introduction

Since its introduction in the 1970s, major advances have taken place in the prenatal detection of structural abnormalities. Several studies have shown the interest of obstetric ultrasound in the prenatal diagnosis of congenital malformations. Indeed, screening ultrasound performed during the three trimesters of pregnancy allows the detection of approximately 60% of serious fetal malformations (1).

In developing countries and in our country,

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routine antenatal screening for malformations is not generalized and there is still no national registry of all cases of congenital malformations.

Prenatal diagnosis is a generic term that includes screening for fetal conditions, their diagnosis, and the practices of appropriate fetal therapy.

Over the past 40 years, the evolution of obstetrics has been marked by the establishment of its three main components which have now become the standard bases of care during pregnancy; thus the first amniocentesis were carried out in the 1970s, punctures of the fetal umbilical cord under ultrasound control and blood transfusions in utero 10 years later, and then the dosage of serum markers in the maternal serum carried out with the aim of screening for fetal aneuploidies (1).

The objective of our study is to describe the experience of the maternity and reproductive health hospital "Les Orangers" in Rabat in the prenatal ultrasound diagnosis of congenital anomalies and to identify the main anomalies detected, their percentage and their short-term outcome.

Methods

Study design

The study took place at the maternity and reproductive health hospital "Les Orangers" in Rabat (HMSRO). This hospital is a level 3 government structure with an average of 8,000 deliveries per year.

This study included all pregnant women whose fetuses or newborns had one or more malformations detected by antenatal ultrasound and/or discovered on clinical examination at birth, regardless of the term or outcome of pregnancy.

The parents were informed about the methods and objectives of the study and their consent was obtained.

Cases of refusal to participate in the survey were excluded as well as cases where the malformation was suspected on obstetric ultrasound but invalidated at birth.

Data collection

The data of the cases of malformations diagnosed on the obstetric ultrasound performed in our maternity were collected and reported on pre-established sheets and on the register of malformations of the hospital.

Definitions

There are three types of ultrasound according

to the CNTEP: (2)

- Systematic or screening ultrasound: includes three systematic examinations during the followup of a normal pregnancy:

• In the first trimester: between 11 and 13 weeks of amenorrhea and 6 days (allows dating, diagnosis of multiple pregnancies, assessment of the risk of chromosomal abnormality, screening for certain malformations);

• In the second trimester: between 20 and 22 weeks of amenorrhea (screening for malformations);

• In the third trimester: between 30 and 32 weeks of amenorrhea (study of fetal growth and screening for certain malformations, location of the placenta).

- Second-line, so-called "diagnostic" ultrasound:

Allows to invalidate or confirm the reality of a fetal pathology, it is indicated when a high risk of fetal morphological abnormality is identified by the anamnesis, or the discovery of an abnormal image during a screening examination. It also helps to clarify the severity of the fetal pathology and to guide practical management.

- Focused ultrasound: performed for specific indications, relating to certain specific points, such as monitoring the amount of amniotic fluid at the end of pregnancy, evaluating fetal "well-being" in the context of monitoring intrauterine growth retardation (Doppler), fetal vitality, presentation, placental location.

Statistical analysis

This is a cross-sectional study. Data entry and statistical analysis were performed using SPSS 18.0 software. Qualitative variables were expressed in numbers and percentages, and quantitative variables in mean and standard deviation.

Ethical approval

The Ethics Committee for Biomedical Research, Faculty of Medicine and Pharmacy – Rabat, University Mohammed V – Rabat, Morocco, approved the study, n°: 20/16.

Results

During the study period, among the 43,923 births, 245 cases of congenital malformations were diagnosed, representing a total of 470 types of congenital malformations over a period of five and a half years. The total prevalence was 5.58 per thousand births (55.78 per 10,000 births).

Prenatal diagnosis by ultrasound was performed in a third of cases, i.e. 33%, thus

revealing 155 congenital anomalies; of which almost half during the 2nd and 3rd trimester with a rate of 51-45% successively, and in 4% during the 1st trimester (Table 1), while in two thirds of cases (67%) the diagnosis was placed at birth

during a systematic clinical examination of the newborn.

The analysis of the socio-demographic characteristics of the cases of congenital malformations diagnosed by antenatal ultrasound

Table1. The different types of congenital anomalies detected on obstetric ultrasound during the 1st trimester, 2nd trimester and 3rd
trimester of pregnancy and at birth at the maternity and reproductive health hospital "Les Orangers" in Rabat between 2011-2016

trimester of pregnancy and at birth at the maternity and reprod Types and subtypes of congenital malformations according to classification (ICD-10)	N		Antenatal d		Postnatal diagnosis	
		T1	T2	Т3	Total	
Q00-Q07 Congenital malformations of the nervous system Anencephaly (Q00) Hydrocephalus (Q03.9) Spina bifida (Q05.9) Encephalocele (Q01.9)	84 22 26 17	2(3.5) 2(11) 0 0	21(37.5) 7(39) 6(28.6) 4(50)	33(59) 9(50) 15(71.4) 4(50)	56(66.6) 18(81.8) 21(80.8) 8(47)	28(33.3) 4(9.1) 5(19.2) 9(53)
Dandy walker Syndrome (Q03.1) Arnold Chiari Syndrome (Q07.0) Microcephaly (Q02) Holoprosencephaly (Q04.2)	5 2 1 4	0 0 0 0	2(66.7) 0 1(100)	1(33.3) 1(100) 1(100) 0	3(60) 1(50) 1(100) 1(25)	2(40) 1(50) 0 3(75)
Absence of Thalami [*] Agenesis of corpus callosum (Q04.0) Facial Paralysis (Q07.8)	4 1 1 1	0 0 0	1(100) 0 0	0 1(100) 1(100)	1(25) 1(100) 1(100) 0	3(75) 0 0 1(100)
Q10–Q18 Congenital malformations of the eye, ear, face & neck Craniofacial dysmorphism* Short neck* Retrognatism* Canines* Low-set ears (Q17.4) Exophtalmia (Q11.3) Microphtalmos (Q11.2) Congenital glaucoma (Q15.0) Frontal bossing* Congenital absence of auricle (Q16.0) Gum tooth*	56 29 6 1 6 3 1 1 1 1 1	0 0 0 0 0 0 0	6(35.3) 4(50) 0 0 1(25) 1(100)	$11(64.7) \\ 4(50) \\ 1(100) \\ 2(100) \\ 1(100) \\ 3(75) \\ 0$	$\begin{array}{c} 17(30.3) \\ 8(27.6) \\ 1(16.7) \\ 2(33.3) \\ 1(100) \\ 4(66.7) \\ 1(33.3) \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ $	$\begin{array}{c} 39(69.6)\\ 21(72.4)\\ 5(83.3)\\ 4(66.7)\\ 0\\ 2(33.3)\\ 2(66.7)\\ 1(100)\\ 1(100)\\ 1(100)\\ 1(100)\\ 1(100)\\ 1(100)\\ 1(100)\end{array}$
Q20–Q28 Congenital malformations of the circulatory system Cardiomegaly* Congenital valve malformation* Cystic hygroma* Single umbilical artery (Q27.0) Hydrothorax* Laevocardia (Q24.1) Tetralogie of Fallot (Q21.3) Anasarca fetoplacental* Cardiomyopathy (Q24.9) hypoplastic umbilical artery (Q27.0) 2 arteries + 2 veins*	38 5 10 5 4 2 1 1 7 1 1 1	2(20) 0 1(25) 0 0 1(100)	4(50) 0 1(100) 3(75) 0 0 0 0 0	$\begin{array}{c} 4(40) \\ 1(100) \\ 0 \\ 1(100) \\ 1(100) \\ 1(100) \\ 0 \end{array}$	$10(26.3) \\ 1(20) \\ 1(10) \\ 4(80) \\ 1(25) \\ 1(50) \\ 1(100) \\ 1(100) \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 \\ 0 $	$28(73.7) \\ 4(80) \\ 9(90) \\ 1(20) \\ 3(75) \\ 1(50) \\ 0 \\ 0 \\ 7(100) \\ 1(100$
Q30–Q34 Congenital malformations of the respiratory system Hypoplasia and dysplasia of lung (Q33.6) Agenesis of lung(Q33.3) Agenesis of nose cartilage (Q30.1)	5 2 1 2	0 0 0	1(50) 1(100) 0	1(50) 0 1(100)	2(40) 1(50) 1(100) 0	3(60) 1(50) 0 2(100)
Q38-Q45 Cleft lip and cleft palate Cleft palate with cleft lip (Q37) Cleft lip(Q36) Cleft palate (Q35) Alveolar cleft* Velopalatine cleft *	21 9 7 3 1 1	0 0 0 0	2(66.7) 1(100) 1(100) 0	1(33.3) 0 0 1(100)	$3(14.3) \\ 1(11.1) \\ 1(14.3) \\ 1(33.3) \\ 0 \\ 0$	18(85.7) 8(89) 6(85.7) 2(66.7) 1(100) 1(100)

Fable 1. Continued 028-045 Congonital malformations of the digestive system						
238-Q45 Congenital malformations of the digestive system	10					10(70 0
Atresia of oesophagus (Q39.0)	18				5(27.8)	13(72.2
Hepatomegaly (Q44.7)	6	0	4(00)		2(33.3)	4(66.7)
mperforate anus (Q42.3)	3	0	4(80)	1(20)	1(33.3)	2(66.7)
Congenital dilatation of the colon (Q43.9)	2	0	2(100)	0	1(50)	1(50)
Anal malformation(Q43.9)	1	0	0	1(100)	1(100)	0
Congenital malformation of mouth NOS (Q38.6)	2	0	1(100)	0	0	2(100)
ligh archedpalate(Q38.5)	1	0	1(100)	0	0	1(100)
Macroglossia(Q38.2)	1				0	1(100)
Glossoptosis *	1				0	1(100)
	1				0	1(100)
250-Q56 Congenital malformations of genital organs						
Typospadias (Q54.9)					6(24)	19(76)
Micropenis (Q55.6)	25				1(14.3)	6(85.7)
Ambiguous genitalia(Q56.4)	7	0	3(50)	3(50)	2(28.6)	5(71.4
Congenital malformation of female genitalia(Q52.9)	7	0	0	1(100)	2(20.0) 2(33.3)	4(66.7)
Cryptorchism (Q53.9)	6	0	0	2(100)	1(100)	4(00.7
nvisible clitoris (Q52.6)	1	0	2(100)	0	1(100)	0
IIVISIBLE CITCOLIS (Q.52.0)	3	0	1(100)	0	0	3(100)
	1				0	1(100)
					0	1(100)
Q60-Q64 Congenital malformations of the urinary system						
Polycystic kidney (Q61.3)	10				7(70)	3(30)
Jreterohydronephrosis (Q62)	2	1(14.3)	3(42.8)	3(42.8)	2(100)	0
Absence of bladder (Q64.5)	2	0	1(50)	1(50)	2(100)	0
Congenital dilatation of ureter (Q62.2)	2	1(50)	1(50)	0	2(100)	0
Epispadias (Q64.0)	1	0	1(50)	1(50)	1(100)	0
Congenital displaced kidney (Q63.2)	2	0	0	1(100)	0	2(100)
	1			(¹)	0	1(100)
Q65–Q79 Congenital malformations and deformations of the						
nusculoskeletal system	15				45(29)	110(71
Clubfoot (Q66.8)	5				4(14.8)	23(85)
Omphalocele (Q79.2)	27				2(16.7)	10(83.3
Accessory fingers (Q69.0)	12				5(45.5)	6(54.5
Reduced limbs (Q73.8)	11				5(50)	5(50)
Gastroschisis (Q79.3)	10	0	33(73.3)	12(26.7)	4(50)	4(50)
Chondrodysplasia punctata (Q77.3)	8	0	3(75)	1(25)	4(66.7)	2(33.3)
Falipes equinovarus (Q66.0)	6	0	1(50)	1(50)	1(20)	4(80)
Macrocephaly (Q75.3)	5	0	4(80)	1(20)	1(20)	4(80)
Syndactyly (Q70)	5	0	5(100)	0	1(25)	3(75)
Гое agenesis*	4	0	4(100)	0	1(25)	3(75)
Clinodactyly*	4	0	2(50)	2(50)	1(33.3)	2(66.7
Congenital anomaly of limb (Q74.9)	3	0	1(100)	0	1(33.3)	2(66.7
Րhanatophoric dysplasia*	3	0	1(100)	0	3(100)	0
Hypertelorism (Q75.2)	3	0	1(100)	0	1(50)	1(50)
Congenital absence of limbs (Q73.0)	2	0	1(100)	0	1(50)	1(50)
limbs in hyperflexion *	2	0	0	1(100)	2(100)	0
Scoliosis (Q67.5)	2	0	1(100)	0	1(50)	1(50)
Diaphragmatic hernia (Q79.0)	2	0	2(66.7)	1(33.3)	1(50)	1(50)
Varrow thorax (Q67.8)	2	0	0	1(100)	1(50)	1(50)
Thin thorax (Q67.7)	2	0	1(100)	0	1(100)	0
Deformed thorax (Q76.9)	1	0	1(50)	1(50)	1(100)	0
Amniotic bands *	1	0	1(100)	0	1(100)	0
Microdactyly*	1	0	1(100)	0	1(100)	0
Spine agenesis *	1	0	1(100)	0	1(100)	0
Prune Belly syndrome (Q79.4)	1	0	0	1(100)	0	5(100)
Feet valgus (Q66.6)	5	0	0	1(100)	0	3(100)
Fingers agenesis*	3	Õ	1(100)	0	0	3(100)
Craniosynostosis (Q75.0)	3	0 0	1(100)	0	0	3(100)
Typerlaxity ligament*	3	0	0	1(100)	0	2(100)
Forearm agenesis*	2	5	5	-(-00)	0	2(100)
	4					
	2				0	21100
Foot agenesis*	2				0	
	2 2 2				0 0 0	2(100) 2(100) 2(100)

Hand agenesis*	2				0	1(100)
Phalanges agenesis*	1				0	1(100)
Thumb hypoplasia*	1				0	1(100)
Club hand*	1				0	1(100)
Bone growth*	1				0	1(100)
Sarcum agenesis*	1				0	1(100)
Paro-occipital bone agenesis*	1				0	1(100)
Asymetric thorax (Q76.9)	1				0	1(100)
Malformation of ribs (Q76.6)	1				0	1(100)
Expanded Thorax (Q67.8)	1				0	1(100)
Congenital funnel chest (Q67.6)	1				0	1(100)
······	1				-	-()
	-					
Q80-Q89 Other congenital malformations	18				3(16.7)	15(83.3)
Situs inversus (Q89.3)	1				1(100)	0
Cyclopia (Q87.0)	1				1(100)	Ő
Ombilical hernia*	1				1(100)	0
Ichtyosis vulgaris (Q80.0)	3				0	3(100)
Cervico-facial hemolymphangioma*	2	0	2(66.7)	1(33.3)	0	2(100)
Congenital splenomegaly*	2	0	0	1(100)	0 0	2(100)
Alopecia (Q84.0)	1	0	1(100)	0	0	1(100)
Naevus (Q82.5)	1	0	1(100)	0	0	1(100)
Epidermal sinus*	1	0	0	0	0	1(100)
Depigmentation*	1			0	0	1(100)
Umbilical cord membrane detachment*	1				0	1(100)
Pierre Robin syndrome (Q87.0)	1				0	1(100)
Acardiac fetus*	1				0	1(100)
Absence of gluteal fold*	1				0	1(100)
Absence of glutear lolu	1				0	1(100)
090–099 Chromosomal abnormalities	40				1(2.5)	39(97.5)
Senior Loken syndrome*	1				1(100)	0
Trisomy 21 (=Down syndrome) (Q90)	35	1(100)			0	35(100)
Trisomy 13 (091.7)	2	1(100)	0	0	0	2(100)
Trisomy 18 (Q91.3)	1	1(100)			0	1(100)
Trisomy 8 (Q92.1)	1				0	1(100)
Total	470	6 (3.9)	79 (51)	70 (45.1)	155 (33)	315(67)
N: Number of malformations %: nercentage T: trimester*						

N: Number of malformations, %: percentage, T: trimester,*: Congenital malformation not found in the ICD-10 classification, ICD-10: International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10)

examination revealed that the mean gestality was 2.46 with extremes of 1 to 8 and the mean parity was 1.69 with extremes of 0 to 6. About 29% of women were primiparous, while 47.8% were pauciparous and 7.2% were multiparous. The average age of the mothers was 29.28 \pm 6.80 with extreme ages of 18 and 46 years. 64% of women are of low socio-economic level. 81.2% of women followed their pregnancy. The average gestational

age was 34.07 ± 6.22 weeks of amenorrhea.

The outcome of the pregnancy was marked by fetal death in utero (FDIU) in 39.1% of cases (Table 2); most of which are represented by polymalformative syndrome in 37% of cases and anencephaly in 33%. Medical termination of pregnancy was performed in 15.9% of cases (11 cases) among all diagnosed cases which is 69; including 36.5% for polymalformative syndrome

Table 2. The outcome of the different types of congenital malformations identified in the maternity and reproductive health hospital "Les

 Orangers" in Rabat between 2011-2016

Types and subtypes of cases of congenital malformations according	N/%		Evolut	ion N/%	
to the international classification ICD-10 diagnosed in antenatal	Total	Good	Death	FDIU	Unknown
Diagnosed polymalformative syndromes	35(50.7)	3(8.5)	17(48.5)	10(28.5)	5(14.5)
Total of isolated CM diagnosed antenatally	34(49.3)	1(2.9)	12(35.3)	17(50)	4(11.8)
Q00-Q07					
Congenital malformations of the nervous system	20(58.8)	1(5)	6(30)	10(50)	3(15)
Anencephaly (Q00)	14(70)	0	5(35.7)	9(64.3)	0
Hydrocephalus (Q03.9)	5(25)	1(20)	0	1(20)	3(60)
Dandy Walker Syndrome (Q03.1)	1(5)	0	1(100)	0	0
Q20–Q28					
Congenital malformations of the circulatory system	7(10.1)	0	2(28.6)	5(71.4)	0
Cystic hygroma*	3(42.9)	0	1(33.3)	2(66.7)	0
Hydrothorax*	1(14.3)	0	1(100)	0	0
Anasarca fetoplacental*	3(42.9)	0	0	3(100)	0

Table 2. Continued					
Q65–Q79 Congenital malformations and deformations of the					
musculoskeletal system	7(10.1)	0	4(57.1)	2(28.6)	1(14.3)
Reduced limbs (Q73.8)	1(14.3)	0	1(100)	0	0
Gastroschisis (Q79.3)	2(28.6)	0	1(50)	0	1(50)
Chondrodysplasia punctata (Q77.3)	1(14.3)	0	1(100)	0	0
Thanatophoric dysplasia*	3(42.9)	0	1(33.3)	2(66.7)	0
Total of CM diagnosed prenatally	69(28.2)	4(5.8)	29(42)	27(39.1)	9(13.1)

N: Number of malformations, %: percentage, CM: congenital malformations, FDIU: fetal death in utero, *: Congenital malformation not found in the ICD-10 classification, ICD-10: International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10)-

(4 cases), 18% for hydrops fetalis (2 cases) and 18% for anencephaly (2 cases), a case of cystic hygroma, a case of senior Loken syndrome, and a case of hydrocephalus associated with an anamnios at 26WA. 68.1% of women gave birth naturally (this rate includes all cases expelled vaginally including medical termination of pregnancy) and 30.4% gave birth by caesarean section. Delivery was premature in 36.2% of cases and was complicated by acute fetal distress in 11.6%. IUGR was described in 14.5% of cases. The short-term evolution of the cases (in the first six months) showed a mortality rate which reaches 42%, this high death rate, is related on the one hand to the severity of the malformation, in particular polymalformations and non-viable congenital malformations, and on the other hand to prematurity and IUGR (Table 3).

The different types of congenital malformations found have been classified according to the international classification (ICD-10) (3).

Table 3. Causes of death of isolated CM diagnosed antenatally at the maternity and reproductive health hospital "Les Orangers" in Rabatbetween 2011-2016

Types and subtypes of cases of isolated congenital malformations (according to the international classification ICD-10) diagnosed antenatally	GA (WA)	Causes of death
Q00-Q07 Congenital malformations of the nervous system		
Anencephaly (Q00)		Non viable
Hydrocephalus (Q03.9)	34	Prematurity
Dandy Walker Syndrome (Q03.1)	37	Respiratory distress
Q20–Q28 Congenital malformations of the circulatory system		
Cystic hygroma*		Non viable
Hydrothorax*	35	Prematurity
Anasarca fetoplacental*		Non viable
Q65–Q79 Congenital malformations and deformations of the musculoskeletal system		
Reduced limbs(Q73.8)	32	Prematurity
Gastroschisis (Q79.3)	34	Prematurity
Chondrodysplasia punctata (Q77.3)	29	Prematurity
Thanatophoric dysplasia*	39	Hypotrophy + Anamnios

GA: gestational age, WA: week of amenorrhea, *: Congenital malformation not found in the ICD-10 classification, ICD-10: International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10)

Discussion

Prenatal ultrasound diagnosis can establish the precise nature of the lesion in 90% of cases without impacting the symptomatology at birth. The most common term for screening for these malformations is the second trimester. Even if it is currently difficult to speak of morphological ultrasound for a given examination, each of the ultrasounds performed during pregnancy must be considered as "morphological". In our context, the antenatal diagnosis was established in a third of the cases (i.e. 33%) of all the abnormalities detected, which corresponds to 28.2% of all the cases highlighted. However, this generally low rate compared to other studies carried out in the world probably does not reflect reality. 19% of women did not follow their pregnancy and the postnatal screening rate reached 67%.

France has implemented a policy of prenatal screening for congenital anomalies by systematizing 3 ultrasounds during the 1st, 2nd and 3rd trimester, and screening for trisomy 21 by serum markers on the one hand, and by measuring nuchal translucency on 1st trimester ultrasound on the other hand. As a result, prenatal diagnosis increased from 16.5% (95% CI 14.1-

19.1) in 1983 to 70.7% (95% CI 68.3-73.1) of malformation cases congenital infections in 2007 (4, 5), and from 20% to 70% according to the High Authority of Health (HAS) (6). Another study described by Munim (7) reports a rate of 48.8%.

Half of our cases (51%) were detected during obstetric ultrasound in the 2nd trimester and 45% during the 3rd trimester. According to Dulgheroff (8), the detection rates of major structural abnormalities in the 1st and 2nd trimester vary from 13% to 43.6% and from 21% to 85% respectively.

Only 6 cases of congenital anomalies (4%) were diagnosed during the 1st trimester in our study. These were Senior Loken syndrome, one case of Ureterohydronephrosis, one case of Tetralogy of Fallot, a cystic hygroma, and two cases of anencephaly.

No case of trisomy 21 was detected prenatally. However, 1st trimester ultrasound has, for 26 years, made it possible to rationalize more efficient and earlier screening for fetal chromosomal abnormalities by standardized and quantitative measurement of the thickness of the nuchal translucency (1), and several studies carried out have shown the performance of 1st trimester ultrasound in the prenatal diagnosis of most birth defects; Cedergren and Selbing found a rate of 41% of anomalies detected between 11-14 WA (9), Weisz (10) showed a rate of 44%. Another study conducted in Finland reported an increase in the detection rate of congenital anomalies in early pregnancy by transvaginal ultrasound from 22% to 79% over six years (11).

A recent study carried out between 2012-2016 noted an abnormality detection rate during the 1st trimester at 27% with a false positive rate of 0.04% (12).

In our study, it is mainly urological abnormalities and malformations of the central nervous system that were screened antenatally with respectively 70% and 67% of cases. This high frequency of these two groups of anomalies was also reported by Munim (7). The other abnormalities involved in antenatal ultrasound diagnosis in our series are malformations of the respiratory system in 40% followed by abnormalities of the musculoskeletal system in 29%, of the digestive system in 28% and a single case of genetic abnormality (2.5%) corresponding to Senior Loken syndrome. According to Bardi et al. (12), the detection rate of chromosomal abnormalities reached 77.7% while in our study 97.5% of cases of chromosomal abnormalities were diagnosed at birth during a systematic clinical examination of the newborn.

Regarding the time of diagnosis, we note that most neurological abnormalities were diagnosed during the 3rd trimester, this finding was also reported by Dulgheroff (8). Similarly, malformations of the eye, ear, face and neck were diagnosed in the 3rd trimester.

On the other hand, malformations of the digestive system, musculoskeletal system, circulatory system and cleft lip and palate were visualized during a 2nd trimester obstetric ultrasound while the detection rate of congenital malformations of the urinary, genital and respiratory systems are identical during the 2nd and 3rd trimester.

As for the evolution of congenital malformations. 42% died either in the immediate postpartum or in the medium term (within 6 months of postpartum); and 39% are FDIU; in the literature, this rate varies between 20 and 30% of perinatal deaths (7). This high death rate is related on the one hand to the severity of the malformation, in particular poly malformations and non-viable congenital malformations, and on the other hand to prematurity, the rate of which reaches 36.2% and IUGR with a rate 14.5% of cases. Only 6% of cases had a good evolution after having had good care in specialized services.

Limitation

This study has some limitations:

-It is a descriptive study carried out in a single center.

- The absence of a national register and the non-computerization of the health system make it difficult to collect data on a large population.

- Poorly structured and often incomplete prenatal follow-up makes information on pregnancy follow-up and prenatal diagnosis unavailable.

- In the event of fetal death in utero, only xrays and autopsy can provide information on the etiology of death; something that is not always accepted by the couple.

- In order to declare all anomalies, including functional anomalies, it is necessary to carry out long-term follow-up, which was not the case in the present study.

Conclusion

Our study allowed us to find weak points in our pregnancy monitoring system; among which:

- A lack of awareness of pregnant women on the need to monitor their pregnancy.

- A low rate of cases of congenital

malformations diagnosed before birth, prompting the establishment of a prenatal screening strategy for congenital malformations generalized throughout the country.

- The lack of screening strategy for Trisomy 21, in particular by measuring nuchal translucency in the 1st trimester of pregnancy.

- The problem of the national register of congenital malformations that must be set up; congenital malformations registries make it possible to measure the impact of prenatal screening policies for congenital anomalies.

- The delay in the implementation of fetal medicine and the training of health professionals in the prenatal ultrasound diagnosis of congenital anomalies.

- Accordingly, a systematized policy within the framework of congenital malformations should be implemented urgently.

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Conflicts of interest

The authors declare that there is no conflict of interest.

References

- 1. Philippe Descamps. Prenatal diagnosis in practice. Elsevier-Masson 2011, 528p.
- Fetal ultrasounds for medical purposes and not medical: definitions and compatibility. Report Technological Assessment. High Authority of Health (HAS)/ 26 April 2012. Available from: https://www.cfef.org/archives/bricabrac/echoHAS. pdf
- 3. World Health Organization, editor. International Statistical Classification of Diseases and related

health problems: Alphabetical index. World Health Organization; 2004. Available from: https://www.atih.sante.fr/sites/default/files/public /content/3069/cim-10_fr_2017.pdf

- 4. Lelong N, Thieulin AC, Vodovar V, Goffinet F, Khoshnood B. Epidemiological surveillance and prenatal diagnosis of congenital anomalies in the Parisian population 1981-2007. Arch Pediatr. 2012;19(10):1030-1038.
- 5. De Vigan C, Khoshnood B, Lhomme A, Vodovar V, Goujard J, Goffinet F. Prevalence and prenatal diagnosis of congenital malformations in the Parisian population: twenty years of surveillance by the Paris Registry of congenital malformations. J Gynecol Obstet BiolReprod 2005;34 (cahier 1): 8-16.
- 6. Gilles Grange. Practical guide to obstetric and gynecological ultrasound. 2nd edition, Elsevier-Masson, 2016.
- 7. Munim S, Nadeem S, Khuwaja NA. The accuracy of ultrasound in the diagnosis of congenital abnormalities. J Pak Med Assoc. 2006;56(1):16-18.
- 8. Dulgheroff FF, Peixoto AB, Petrini CG, Caldas TMRDC, Ramos DR, Magalhães FO, et al. Fetal structural anomalies diagnosed during the first, second and third trimesters of pregnancy using ultrasonography: a retrospective cohort study. Sao Paulo Med J. 2019;137(5):391-400.
- 9. Cedergren M, Selbing A. Detection of fetal structural abnormalities by an 11-14-week ultrasound dating scan in an unselected Swedish population. Acta Obstet Gynecol Scand. 2006;85(8):912-915.
- Weisz B, Pajkrt E, Jauniaux E. Early detection of fetal structural abnormalities. Reprod Biomed Online. 2005;10(4):541-553.
- 11. Taipale P, Ammälä M, Salonen R, Hiilesmaa V. Learning curve in ultrasonographic screening for selected fetal structural anomalies in early pregnancy. Obstet Gynecol. 2003;101(2):273-278.
- Bardi F, Smith E, Kuilman M, Snijders RJM, Bilardo CM. Early Detection of Structural Anomalies in a Primary Care Setting in the Netherlands. Fetal Diagn Ther. 2019;46(1):12-19.