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SYNDROMES ASSOCIATED WITH SINGLE UMBILICAL ARTERY

Pathology

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ABSTRACT

Introduction: The umbilical cord connects the fetus to the placenta at the feto-maternal interface and typically consists of two umbilical arteries and one umbilical vein. However, in some cases, only a single umbilical artery (SUA) is present. As single umbilical artery is associated with poor perinatal outcome and association of other fetal anomalies, a detailed evaluation of the fetus for other anomalies is warranted. This study aimed to study the syndromes associated with SUA identified during perinatal autopsies. Aims and objectives: To study syndromes associated with single umbilical artery in perinatal autopsies. **Materials And Methods:** We reviewed the records of all fetuses submitted for autopsy to the Department of Pathology over a 5-year period (2019–2024), totalling 503 cases. The data were retrieved from the hospital's pathology laboratory records. Autopsy specimen were examined for single umbilical artery and syndromes associated with single umbilical artery were noted. Anthropometry was recorded, X-ray, CT-Scan and MRI were reviewed where ever necessary. Rokitansky technique of en block dissection done. **Results:** A SUA was present in 12.3% of the autopsies. The gestational age of the affected fetuses ranged between 20 weeks to 7 days after birth. 10 cases (2%) of single umbilical artery were associated with syndromes namely 3 cases of sirenomelia, 2 cases of OEIS complex, 1 case each of Dandy Walker Syndrome, Arnold Chiari Malformation, Jacobsen syndrome, Potters syndrome, Edward's syndrome. **Conclusion:** A SUA is usually seen in association with other syndromes. Perinatal autopsy plays a crucial role in understanding the implications and underlying causes associated with a single umbilical artery (SUA).

KEYWORDS

Syndromes, Perinatal autopsy, Single umbilical artery.

INTRODUCTION

Single umbilical artery (SUA) is one of the common congenital anomalies with a reported incidence of 0.2-1.2% [1]. About 33% of fetuses with single umbilical artery have associated structural anomalies and 10 % have chromosomal anomalies.[1]

It is commonly associated with: Monozygous twins, Sirenomelia, VACTERL anomalies, Fetal hydantoin syndrome, Meckel–Gruber syndrome, Jarcho–Levin syndrome, Multiple lentigines syndrome, Zellweger syndrome, Trisomies 13 and 18 and rarely Trisomy 21 and Monosomy 45X.[2]

Single Umbilical Artery (SUA) is the most common true congenital anomaly of the umbilical cord and was first described by Vesalius [3].

When compared with congenital anomalies of all other systems, SUA has a prevalence rate of 1% [4].

Normally, the umbilical cord contains two arteries and one vein. SUA is a condition characterised by the presence of only one umbilical artery. This anomaly is caused by developmental agenesis of one umbilical artery, marked hypoplasia of a previously normally developed umbilical artery, or persistent allantoic artery of the body stalk [5].

The incidence of SUA in twins is three times that of singletons[6].

Previous studies in the literature, depending on the target population being studied, including autopsies, prenatal ultrasonographic examination, or in foetuses born alive or preterm, have reported different rates of SUA detection[7]

As single umbilical artery is associated with poor perinatal outcome and association of other fetal anomalies is not clearly understood, a detailed evaluation of the fetus for other anomalies is warranted.[4]

The perinatal autopsy can provide important information to the family, the clinician, and society. In cases where the pregnancy was terminated due to malformations, post mortem can confirm, modify, or exclude a prenatal diagnosis, i.e., it serves as an audit tool for both diagnosis and diagnostic techniques. [8]

The present study was therefore undertaken with the objective to determine the syndromes associated with single umbilical artery.

Thus inspite of modern antenatal diagnostic modalities, perinatal autopsy plays an important role in confirming the diagnosis, delineating a spectrum of anomalies and helps in embryological understanding.

MATERIALS AND METHODS

We reviewed the records of all fetuses submitted for autopsy in the Department of Pathology over a 5-year period (2019–2024), totaling 503 cases. These fetuses were either aborted due to intrauterine death or after prenatal detection of a malformation. The data were retrieved from the hospital's pathology laboratory records. Autopsy specimen were examined for single umbilical artery and syndromes associated with single umbilical artery were noted. Anthropometry was recorded, X-ray, CT-Scan and MRI were reviewed where ever necessary. Rokitansky technique of en block dissection done.

RESULTS

We examined 503 fetuses and the gestational age ranged between 20 weeks to 7 days after birth. and the birth weight between 150g and 4000 g. Single umbilical artery was found in 62 cases (12.3%). Out of these 62 cases, 10 cases (16.1%) were associated with syndromes. Among these there were 3 cases (4.8%) of sirenomelia **(Fig1:1A)** , 2 cases (3.2%) of OEIS complex **(Fig 2: 2A,2B)**, 1 case(1.6%) each of Dandy Walker Syndrome, Arnold Chiari Malformation**(Fig 3: 3A),** Jacobsen syndrome, Potters syndrome, Edward's syndrome **(Fig 4: 4A,4B)** as shown in **Table 1**

Table 1: Syndromes Associated With Single Umbilical Artery:

Sl. No	Syndrome	Associated anomalies	Number of cases (%)
1	Sirenomelia or Mermaid syndrome	Bilateral renal agenesis, multicystic renal dysplasia, kyphoscoliosis and ambiguous genitalia, and single umbilical artery	3 (4.8%)

2	Dandy Walker Syndrome	Corpus callosum agenesis and single umbilical artery	1(1.6%)
3	Arnold Chiari Malformation	Meningocele, diaphragmatic hernia, bilateral club foot, single umbilical artery	1(1.6%)
4	OEIS Complex	Omphalocele, bladder exstrophy, imperforate anus, kyphoscoliosis and single umbilical artery	2(3.2%)
5	Jacobsen syndrome	Low set ears, rocker bottom feet, hypertelorism, depressed nasal bridge, bilateral lungs showing two lobes , VSD , absent gall bladder and single umbilical artery	1(1.6%)
6	Potters syndrome	Sirenomelia, renal agenesis, absent ureter, urinary bladder, heart shows VSD and single umbilical artery	1(1.6%)
8	Edward's syndrome	Increased head circumference, face-depressed nose, low set ears, micrognathia, hands- bilateral camptodactyly, hyperextended right leg, bilateral rocker bottom foot, interhemispheric cyst.	1(1.6%)
TOTAL			10(16.1%)

DISCUSSION:

Single umbilical artery is the most frequent anomaly, not only of the umbilical cord but probably among all birth defects.

The rate of single umbilical artery showed a 10-fold increase when other malformations were present. The risk for other malformations increased significantly, by a 3-fold to 9-fold measure, when a single umbilical artery was present[9]

In the present study, 503 fetuses were examined and the gestational age ranged between 20 weeks to 7 days after birth. The birth weight ranged between 150g and 4000 g. The incidence of single umbilical artery was 12.3%, as shown in **Table 2.**

Table 2 : Incidence Of Single Umbilical Artery Compared With Other Studies

Study	Autopsies	Single Umbilical Artery	Percentage
Shalini Nayak et al [4](2010)	214	17	7.9%
Mounika P et al [10] (2021)	81	9	11.1%
Chandrahas Kotian et al [6] (2021)	1338	63	4.7%
Present study	503	62	12.3%

Incidence of single umbilical artery in perinatal autopsies in the present study is 12.3% which closely correlated with study by Mounika P et et al[10] showing an incidence of 11.1%, as shown in **table 2.**

Table 3: Association Of Single Umbilical Artery With Syndromes

Study	Total number of SUA cases	Association with syndromes
Chandrahas Kotian et al [6] (2021) (1338 autopsies)	63	6 (9.5%)
Present study (503 autopsies)	62	10 (16.1%)

In the present study, 10 cases (16.1%) of single umbilical artery in perinatal autopsies were associated with syndromes, while in the study by Chandrahas Kotian et al [6] 6 cases (9.5%) of single umbilical artery were associated with syndromes. **(Table 3)**

Among all the syndromes, sirenomelia is the most common syndrome encountered that is 3 cases (4.8%) as shown in while study done by Chandrahas et al[6] showed VATER (4 cases) as Table1 the most common syndrome.

Mermaid or Sirenomelia is a congenital structural anomaly characterized by an abnormal development of the caudal region of the body with different degrees of fusion of the lower extremities. The diagnostic triad suggested by Raabe et al for Sirenomelia /mermaids syndrome consists of (i) fused lower extremities (ii) bilateral renal agenesis (iii) oligohydramnios.[11]

In the present study there were 2 cases of OEIS Complex which were associated with Omphalocele, bladder exstrophy, imperforate anus, kyphoscoliosis and single umbilical artery. It results from improper closure of ventral abdominal wall due to failure of cephalo-caudal and lateral foldings with associated defects of cloaca and urorectal septum. It usually occurs during early blastogenesis around 4th week of development. Other associated anomalies usually associated include cardiovascular, central nervous system, vertebral, upper urinary tract, malrotation, lower extremity anomalies, double appendix, absent appendix, small bowel atresia and abdominal musculature deficiency.[12]

Other rare syndromes encountered in the study were 1 case (1.6%) each of Dandy Walker Syndrome, Arnold Chiari Malformation**,** Jacobsen syndrome, Potters syndrome and Edward's syndrome.

CONCLUSION:

A SUA was present in 12.3% of the autopsies. A SUA is usually seen in association with other syndromes. Perinatal autopsy plays a crucial role in understanding the implications and underlying causes associated with a single umbilical artery (SUA). It provides essential information that can aid in clinical decision-making, genetic counseling, and future medical research. Recent advancement in imaging studies, genetic and molecular techniques can complement but cannot replace a complete and thorough autopsy examination.

Fig 1a: Sirenomelia Type VII:Gross image showing fused lower limbs and X- RAY showing single femur and absent tibia (Sirenomelia type VII)

Other associated anomalies were imperforate anus, kyphoscoliosis, Renal agenesis, Umbilical cord- single umbilical artery

Fig 2A: OEIS Complex: Fetus with Omphalocele, bladder exstrophy

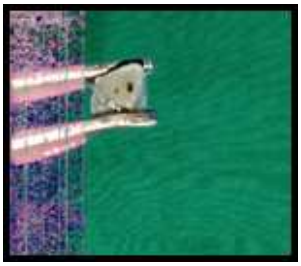

Fig 2B: OEIS Complex: Grossly: Umbilical cord- single umbilical artery
Other anomalies noted were Spinal defect and Imperforate anus

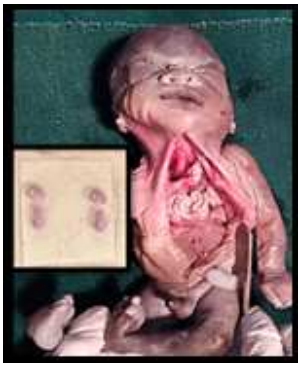

Fig 3A: Arnold Chiari Malformation: Diaphragmatic hernia (stomach and intestine present in thorax) , bilateral club-foot and inset showing Umbilical cord with single umbilical artery.
Other findings noted were- meningocele

Fig 4A: Edward's syndrome: Gross- increased head circumference, face- depressed nose, low set ears, micrognathia, bilateral hands-camptodactyly, hyperextended right leg, bilateral rocker bottom foot.

Fig 4B: Edward's syndrome: Gross-Interhemispheric cyst and inset shows radiologically- interhemispheric cyst noted in the roof of 3rd ventricle measuring 36x30 cm.
Associated with single umbilical artery.

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