Single umbilical artery with associated anomalies at perinatal autopsy

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Abstract

Introduction: Single umbilical artery (SUA) is one of the congenital anomalies, with a birth incidence of about 0.2-1.2%. It may accompany other abnormalities or occur as an isolated defect. The aim of the study was to estimate the association between SUA and other birth defects in a series of perinatal autopsies.

Materials and Methods: We evaluated 322 fetuses sent for autopsy from June 2016 to December 2017 at M.R. Medical College, Kalaburagi.

Results: Of 322, ten fetuses (3.12%) had single umbilical artery. Among which 6 cases were associated with urinary anomalies, 1 case with associated cardiac anomaly, 1 case of inencephaly and 2 were of isolated SUA defect.

Conclusion: Single umbilical artery is commonly associated with other congenital anomalies and it has a strong association with urinary anomalies.

Keywords: Single umbilical artery, Perinatal autopsy, Congenital anomalies, Urinary anomalies.

Introduction

Single umbilical artery is one of the common congenital anomalies with a reported incidence of 0.2-1.2% in live newborns.[1] The incidence is higher among twins, fetal deaths, abortuses and autopsies.[2,3] The rate of associated congenital anomalies with single umbilical artery is about 10% as reported by a National Registry.[4]

Single umbilical artery is detectable by ultrasound after about 13 gestational weeks, and it can be found in healthy newborns, but its prevalence is 3 to 4 times higher in infants with fetal growth restriction, prematurity, perinatal death, and twinning.[3] It has also been associated with maternal diabetes, preeclampsia, and poli- and oligohydramnios,[1,2] and its prevalence is about 4 times higher in autopsy reports[3] or when other birth defectss[2,4–6] are present. However, it is important to look for other associated anomalies when single umbilical artery is diagnosed antenatally or postnatally.

SUA, occurring as a solitary malformation, is compatible with postnatal life.[1] However, the increased incidence of SUA in low birth weight infants suggests that this anomaly exerts a deleterious effect on the intra- uterine growth of the fetus.[1,2] Although SUA may coexist with a variety of abnormalities, it has been found particularly in association with malformations of the renal, skeletal, gastrointestinal, cardiovascular, and central nervous systems.[5-8]

We here present observations on the types of congenital malformations found in association with SUA on perinatal autopsies.

Materials and Methods

We examined 322 fetuses which were sent for autopsy from June 2016 to December 2017, at M.R. Medical College, Kalaburagi. These fetuses were either aborted due to intrauterine death or after prenatal detection of a malformation. The gestational age of the fetuses ranged from 14 to 38 weeks.

Proportions were compared using a chi-square test. The risk for each selected birth defect associated with SUA was expressed in terms of the odds ratio (OR) and its 95% confidence interval (95% CI). All analyses were performed with computer software SPSS 22.0 version.

Result

We examined 322 fetuses and the gestational age ranged between 14 and 38 weeks and the birth weight between 150g and 3400 g. We found single umbilical artery in 3.1% (Table 1) of the autopsies (10 cases) and as a single anomaly in 0.6% (2 cases). Out of these 10 cases, 8 cases were associated with other congenital anomalies. Majority of the cases (i.e 6 cases) were associated with urinary anomalies (Fig. 1 & 2), 1 case of associated inencephaly (Fig. 3) and 1 case of cardiac anomaly (Fig. 4) as shown in table 2. Among these there were also 2 cases of mermaid syndrome or syrenomelia with SUA (Fig. 5).

The overall birth defect prevalence in association with SUA was 80% ((OR 20.47; 95% CI, 4.2-99.2; P <0.001). Urinary tract anomalies predominated among the former, followed by cardiovascular and CNS defects. A preferential association with SUA was only found for urinary tract anomalies (OR 57; 95% CI; P <0.001).

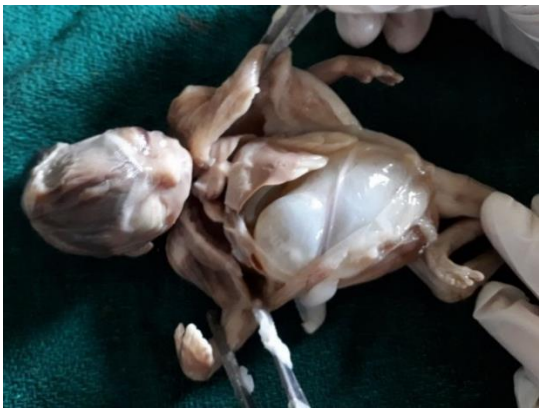

Fig. 1: Gross picture of fetus with megacystis

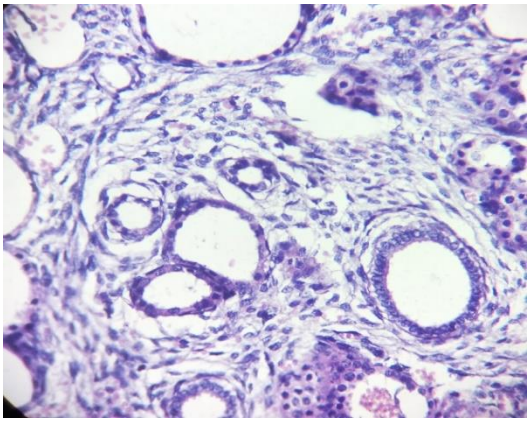

Fig. 2: H & E 40x; showing primitive tubules & ducts lined by cuboidal epithelium surrounded by mesenchymal tissue

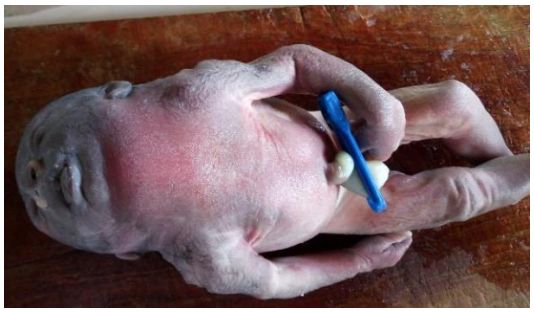

Fig. 3: Gross picture of inencephaly

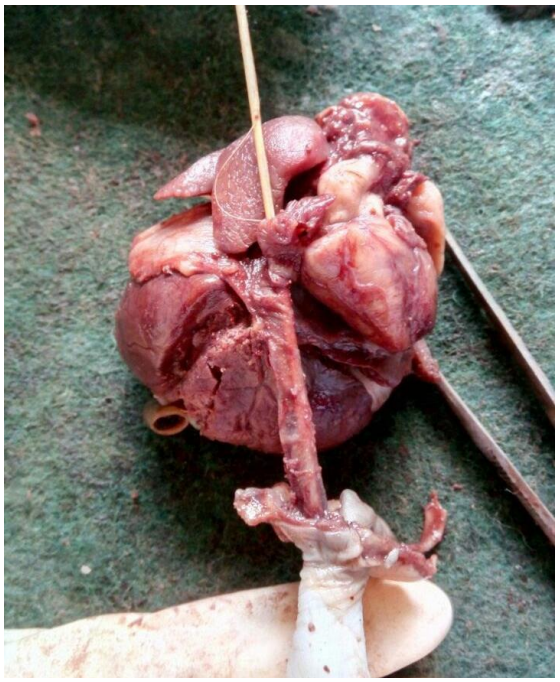

Fig. 4: Gross picture showing absent ductus venosus

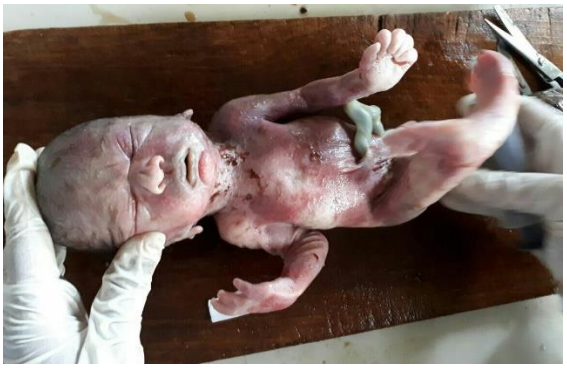

Fig. 5: Gross picture of Mermaid syndrome

Table 1: Showing details of cases with congenital anomalies

Congenital anomalies	With SUA	Without SUA
Nervous system anomalies	01	24
Urinary anomalies	06	08
Lung anomalies	-	08
Cardiovascular anomalies	01	03
GIT anomalies	-	04
Musculoskeletal anomalies	-	04
Total	**08**	**51**

(SUA: Single Umbilical artery)

Table 2: Showing details of all 10 cases of SUA.

S. No	Gestational age	Birth weight	Mode of death	Associated anomalies
1.	32wks	1200g	IUD	Left renal agenesis with B/L CCAM type-2 and diaphragmatic hernia
2.	28wks	1000g	IUD	Mermaid syndrome with B/L renal agenesis
3.	26wks	900g	IUD	Renal-Hepatic-Pancreatic dysplasia (RHPD) with asplenia
4.	27wks	1000g	IUD	Inencephaly
5.	14wks	100g	IUD	Left renal hypoplasia with ventriculomegaly
6.	38wks	3500g	IUD	-
7.	28wks	1500g	Termination	Absent ductus venosus with overriding of aorta, right ventricular hypertrophy and pulmonary stenosis
8.	22wks	1200g	Termination	Mermaid syndrome with left multicystic dysplastic kidney and right renal agenesis
9.	14wks	300g	Termination	Megacystis
10.	28wks	1000g	IUD	-

Discussion

Perinatal mortality of SUA infants is related to associated fetal malformations, prematurity, low birthweight and intra-uterine growth retardation. Although associated malformations are the primary cause of the high perinatal mortality, even non-malformed SUA infants have an increased mortality rate.[4]

It is clear from the literature that SUA is more often found in association with a wide range of other malformations than as an isolated defect, but there is no agreement as to whether preferential associations with particular types of defects exist.[3]

The reported incidence in the literature of congenital malformations in SUA infants vanes between 18.4% and 68%, in prospective studies. In our study the incidence was 80%. It was well correlated with study by Rittler et al[3] Eighty-three percent of SUA cases involved other malformations and 81% reported by Heifetz[6] in an autopsy series but much higher than the 53% reported by Froehlich and Fujikura,[5] probably because of methodological differences.

Peckham and Yerushalmy's[8] prospective study also included a control group; they found that the incidence of congenital malformations in children with three vessels was 1.15% and in SUA infants 20.4%.[4] In our study the incidence in control group is 15.8% and in SUA infants is 80%. In a study by Lilja et al[4] found that the overall risk of malformation in SUA infants is increased 4.3 times. In our study we found the risk to be increased 20.4 times.

In a study by Rittler et al,[3] excluding bilateral kidney agenesis and other severe urinary tract anomalies, close to 10% of the cases with SUA still showed a mild to moderate urinary defect, which was probably not the primary cause of death. The present results showed that 60% of the cases involving SUA had a urinary anomaly of varying degrees of severity and that the risk for this type of birth defects was 57 times higher if SUA was present than if it was absent.

Conclusion

Single umbilical artery has incidence of 3.1%. It is commonly associated with other congenital anomalies and it has a strong association with urinary anomalies.

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How to cite this article: Anita AM, Pattar PM, Patil AG, Savale J, Kamalapurkar M. Single umbilical artery with associated anomalies at perinatal autopsy. J Diagn Pathol Oncol. 2018;4(3):177-179.