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Rarely Encountered Umbilical Cord Pathologies (Literature Review)

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Abstract: This review systematizes current knowledge about umbilical cord abnormalities (UCAs) — a heterogeneous group of anomalies including length abnormalities, number of vessels, attachments, nodes, cysts, and vascular malformations. Based on the analysis of scientific literature (2000–2024), the epidemiological data are presented: UCAs occur in 15–35% of pregnancies, with single umbilical artery (SUA) — in 0.5–5%, velmen insertion — up to 50% in monochorionic twins, and true nodes — in 0.3–1.2%. The review details complications: fetal growth restriction (FGR), chronic and acute hypoxia, antenatal death, risks during childbirth (vascular rupture in vasa praevia, asphyxia). Particular attention is paid to early diagnostic methods: ultrasound with Doppler (blood flow assessment, EAP), color Doppler mapping (Velmen attachment), 3D/4D ultrasound in STIC mode (nodes, spatial anomalies), CTG monitoring. Differentiated management tactics are substantiated: from dynamic observation at low risk to planned cesarean section for vasa praevia or IUGR. It is emphasized that timely diagnostics and an individualized approach reduce perinatal mortality and improve outcomes.

Keywords: Umbilical cord cysts; varicose umbilical veins (varix); Pregnancy complications; Perinatal outcomes; Fetal growth restriction (FGR); Cord structure pathology.

Introduction: In addition to relatively common anomalies (entanglement, EAP), there are rare umbilical cord pathologies that pose significant diagnostic and clinical challenges due to their low incidence, insufficient study, and potentially catastrophic consequences for the fetus. These include umbilical cord hypoplasia, varicose umbilical veins (varix), and true umbilical cord cysts. Their prevalence does not exceed 1-3%, which complicates the accumulation of evidence [1, 2].

The objective of the review is to systematize data on epidemiology, classification (ICD-10/ICD-11), etiopathogenesis, impact on pregnancy and fetus, diagnostic methods, and principles of patient management with these rare pathologies based on an analysis of modern literature (2014-2024).

METHODS

A systematic search was conducted in PubMed, MEDLINE, Scopus, Cochrane Library, and eLibrary databases for the period 2014–2024. Key words: "thin umbilical cord", "umbilical cord hypoplasia", "umbilical vein varix", "fetal intra-abdominal umbilical vein varix", "umbilical cord cyst", "rare umbilical cord abnormalities", "prenatal diagnosis", "pregnancy outcome", "fetal complications", "ICD-10", "ICD-11". Original studies (prospective and retrospective cohort studies, case series), systematic reviews, meta-analyses, and clinical guidelines were included. Publications in languages other than English and Russian, without an abstract, and articles with a low level of evidence (reports of single cases without literature analysis, expert opinions without data) were excluded.

RESULTS

Rare umbilical cord pathologies are characterized by low prevalence. Hypoplasia of the umbilical cord (diameter < 8–10 mm in the second trimester or < 5–8 percentile) occurs in less than 1% of pregnancies and is coded in ICD-10 as O69.8, and in ICD-11 as JA20.2 [1,3,4]. Umbilical vein varices (UVV) - focal enlargement of the vein > 9 mm or > 50% of adjacent areas - is diagnosed in 0.4–1.1% of cases (ICD-10: O69.8/Q27.8; ICD-11: JA20.2/LA8F) [2,5,6]. True umbilical cord cysts

(anechoic formations of embryonic origin) are detected in 0.1–0.5% of cases (ICD-10: O69.8/Q79.8; ICD-11: JA20.2/LB11) [7,8]. Hypoplasia is associated with Wharton's jelly deficiency caused by impaired angiogenesis (decreased VEGF, PlGF) and mesenchymal differentiation. Key risk factors: maternal smoking, thrombophilia, history of IUGR [1,3,4]. UVV develops due to weakness of the venous wall, local elastin defects, increased venous pressure (fetal heart failure, arteriovenous shunts) or portal system thrombosis. Pathogenesis includes turbulent blood flow → endothelial damage → risk of thrombosis/rupture [2,5,6]. True cysts are formed due to the persistence of embryonic structures: the allantois (urogynectic cysts) or the omphalo-mesenteric duct [7,8].

Perinatal risks of rare umbilical cord pathologies are characterized by significant variability. In case of umbilical cord hypoplasia, there is a 3.5–5-fold increase in the risk of fetal growth restriction (FGR) (relative risk [RR] 3.5–5.0), which correlates with chronic hypoxia, manifested by an increase in the resistance index (RI) in the umbilical artery by 40% compared to the norm, as well as an increase in the risk of antenatal fetal death by 2–3 times [1,3,4]. Varicose umbilical vein (UVV) is associated with antenatal death in 8–15% of cases, mainly due to thrombosis, while the risk of intrapartum rupture of the varicose node reaches 4–7% with fetal mortality exceeding 90% [2,5,6,13]. This pathology is also associated with the development of IUGR in 24% of fetuses and a combination with congenital anomalies in 18-30% of cases [5,6]. True umbilical cord cysts diagnosed in the first trimester demonstrate an association with chromosomal abnormalities (mainly trisomy 18) in 15-20% of cases, while large cysts (>40-50 mm) cause vascular compression leading to IUGR in 12-18% of fetuses [7,8,14].

Diagnostic algorithms are based on a comprehensive ultrasound examination. The key criteria for hypoplasia are a decrease in the umbilical cord diameter to less than 8 mm in the second trimester, a decrease in the volume of Wharton's jelly and pathological Doppler parameters (RI>0.8 in the umbilical artery, reverse blood flow in the venous duct), which are predictors of IUGR [1,4]. UVV diagnostics is based on the detection of vein dilation greater than 9 mm in diameter, registration of turbulent flow during color Doppler mapping (CDM) and an increase in peak systolic velocity (PSV) greater than 40 cm/s, which is associated with a high risk of thrombus formation [2,5,6]. True cysts are identified as anechoic formations without internal blood flow during CDM, while the use of three-dimensional ultrasound (3D-ultrasound) allows them to be differentiated from false cysts due to visualization of the epithelial lining [7,8]. As additional methods, fetal magnetic resonance imaging (MRI) demonstrates high sensitivity (92%) in assessing portal system thrombosis in UVV and vascular compression by large cysts (>30 mm) [2,8], and invasive prenatal diagnostics (karyotyping/chromosomal microarray analysis) is recommended for UVV and first trimester cysts due to the frequency of aneuploidies reaching 18% [5,7].

Modern scientific developments (2020–2024) are aimed at improving prognosis and diagnostics. A multiparameter risk index for UVV has been developed, including vein diameter, PSV, and the presence of a thrombus, with an area under the ROC curve (AUC) of 0.89 [6], and for the early detection of complications in hypoplasia, a prognostic significance of a decrease in the umbilical cord diameter to less than 2.5 mm in the first trimester, associated with the development of IUGR in 80% of cases, has been established [3,4]. Technological innovations include the use of 3D power doppler for quantitative assessment of umbilical cord vascularization, where a decrease in the vascularization index correlates with the severity of IUGR in hypoplasia (correlation coefficient r = 0.72) [4], as well as the use of the STIC mode in 4D ultrasound, which increases the accuracy of diagnosing vascular compression by cysts by 35% [8]. In the field of biomarkers, a promising direction is the determination of the sFlt-1/PlGF ratio (>38) in UVV as an indicator of endothelial dysfunction [6], and the detection of elevated levels of alpha-fetoprotein (AFP) and beta-subunit of human chorionic gonadotropin (β-hCG) in amniotic fluid in cysts associated with trisomy 18 [7].

Differentiated tactics are based on the pathogenetic features of each nosology. In case of hypoplasia, intensive monitoring is required (Dopplerometry 2 times a week, CTG from 28 weeks), and the development of blood flow disorders requires early delivery by cesarean section at 34–37 weeks [1,4]. Management of UVV includes planned operative delivery at 34–36 weeks to prevent thrombosis and rupture, supplemented by daily CTG monitoring and weekly Doppler [2,5,6]. For large true cysts (>50 mm), delivery at 36–38 weeks is recommended if there are signs of vascular compression or IUGR [7,8].

Table 1

Principles of management

Pathology	Monitoring	Delivery
Hypoplasia	Ultrasound + Doppler/CTG 2 times a week; hospitalization for IUGR	Planned CS at 34–37 weeks with impaired blood flow
UVV	Ultrasound + Doppler 2 times a week; CTG daily; hospitalization	Planned CS at 34–36 weeks (standard)
cysts > **50 MM**	Growth monitoring Doppler (ultrasound 1 time/2 weeks);	CS at 36–38 weeks with vascular compression

> These approaches are based on cohort studies and systematic reviews of the last decade, emphasizing the need for personalized risk management [1–8,14].

CONCLUSION

Rare umbilical cord pathologies (hypoplasia, umbilical vein varices, true cysts), although occurring in less than 1-3% of cases, are significant causes of perinatal morbidity and mortality. Their timely prenatal diagnosis using modern ultrasound technologies (including Doppler and 3D/4D) and fetal MRI is critically important. The pathogenesis of many of these conditions requires further study. Pregnancy management should be based on a thorough assessment of fetal risks using all available diagnostic tools. In cases of UVV and progressive hypoplasia with IUGR, planned early delivery by cesarean section is indicated. Further studies should be aimed at identifying biomarkers, clarifying the genetic basis and developing standardized protocols for monitoring and delivery to improve perinatal outcomes.

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