



## Original Article

## Prenatal diagnosis of umbilical cord cyst: Clinical significance and prognosis



Leyre Ruiz Campo <sup>a,\*</sup>, Ricardo Savirón Cornudella <sup>a</sup>, Francisco Gámez Alderete <sup>c</sup>,  
Cristina Martínez-Payo <sup>b</sup>, Pilar Pérez Pérez <sup>a</sup>, Pilar Garrido Fernández <sup>a</sup>,  
Diego Lerma Puertas <sup>a</sup>

<sup>a</sup> Department of Obstetrics and Gynecology, Hospital Universitario Miguel Servet, Zaragoza, Spain

<sup>b</sup> Department of Obstetrics and Gynecology, Hospital Universitario Puerta de Hierro, Madrid, Spain

<sup>c</sup> Department of Obstetrics and Gynecology, Hospital Universitario Gregorio Marañón, Madrid, Spain

## ARTICLE INFO

Article history:  
Accepted 30 June 2016

**Keywords:**  
Cystic  
Pregnancy outcome  
Ultrasound  
Embryological cord  
Embryological cyst

## ABSTRACT

**Objective:** Clarify the prognosis of the prenatal ultrasound diagnosis of umbilical cord cysts at any gestation trimester and to assess the ultrasound findings and chromosomal alterations associated to this entity.

**Materials and methods:** Between 2003 and 2015 a multicenter study was carried out, collecting, in five centers in Spain, the associated findings and perinatal outcomes of 27 cases of umbilical cord cysts, regardless of gestational age at diagnosis. A bibliographic review was conducted to identify previous studies in order to compare them with our data.

**Results:** In our sample, the prognosis of this finding and the neonatal outcome, when isolated, is favorable, regardless of gestational age at diagnosis, multiple or unique presentation or vanishing or persistent cysts.

**Conclusions:** It is important to properly assess the umbilical cord cyst and when is diagnosed, it is recommended to conduct a meticulous ultrasound examination searching for other associated malformations. In our study the prognosis of this finding seems to be favorable when isolated. Also, there is no relation between prognosis and gestation weeks at diagnosis. On the other hand, when we find this entity with associated anomalies, it is recommended to assess the need to carry out a karyotype.

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## Introduction

Although umbilical cord cysts are a relatively common, they cannot be considered a non-pathological ultrasound finding. Some reviews indicate that small cysts can be found in up to 3% of pregnancies in the first trimester. Sometimes are associated with other structural and/or chromosomal malformations (even up to 20%), [1–7]. If they are persistent and progressive in size, they can compromise the fetus by restricting blood flow through the umbilical cord, either by compression or thrombosis [1,8].

\* Corresponding author. Department of Obstetrics and Gynecology, Hospital Universitario Miguel Servet C, Isabel la Católica, 1-3, 50009 Zaragoza, Spain.

E-mail addresses: [leyre.ruizcampo@huc.es](mailto:leyre.ruizcampo@huc.es) (L. Ruiz Campo), [rsaviron@gmail.com](mailto:rsaviron@gmail.com) (R. Savirón Cornudella), [fgamez@huc.es](mailto:fgamez@huc.es) (F. Gámez Alderete), [cmartinezp@huc.es](mailto:cmartinezp@huc.es) (C. Martínez-Payo), [pperezp@huc.es](mailto:pperezp@huc.es) (P. Pérez Pérez), [pgarrido@huc.es](mailto:pgarrido@huc.es) (P. Garrido Fernández), [lerma@huc.es](mailto:lerma@huc.es) (D. Lerma Puertas).

<http://dx.doi.org/10.1016/j.tjog.2017.08.008>

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With the widespread use of the ultrasound for prenatal control (for example, in Spain for low-risk pregnancies, three ultrasound scans are performed during pregnancy), intrauterine abnormalities involving the umbilical cord are routinely diagnosed, being umbilical cysts the second most frequent disorder founded [9].

This is the first work published so far in which data has been collected and compared from cases diagnosed in any of the gestation trimesters, trying to clarify the prognosis associated to prenatal diagnosis of this entity. Also, the ultrasound findings more frequently associated to this entity and related to chromosomal alterations are described for a better management during pregnancy.

## Material and methods

**IRB:** CP03/2015. The study was conducted between 2003 and 2015 involving five Spanish centers. Three public hospitals, Miguel Servet University Hospital in Zaragoza and Gregorio Marañón

# Prenatal Diagnosis And Prognosis

**Marco Cascella**



## **Prenatal Diagnosis And Prognosis:**

**Circulating Nucleic Acids in Early Diagnosis, Prognosis and Treatment Monitoring** Peter B. Gahan, 2014-11-18  
DNA and RNA fractions can be isolated from a variety of body fluids including whole blood serum plasma urine saliva and cerebrospinal fluid from both patients and healthy individuals Such isolates can be exploited in the early detection of clinical disorders stratification of patients for treatment treatment monitoring and clinical follow up In addition the use in fetal medicine allows the early detection of fetal sex Rh factor and aneuploid disorders as well as following both fetal and premature born infant development This volume is intended as a primer for those who are interested in entering the field of circulating nucleic acids The areas covered in this volume include Background and general biology of circulating nucleic acids Methodology Applications of circulating nucleic acids Quality Assurance Ethics Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases Nenad Blau, Carlo Dionisi Vici, Carlos R. Ferreira, Christine Vianey-Saban, Clara D. M. van Karnebeek, 2022-02-21 This updated and enlarged second edition is a unique source of information on the diagnosis treatment and follow up of metabolic diseases The clinical and laboratory data characteristic of rare metabolic conditions can be bewildering for clinicians and laboratory personnel alike reference laboratory data is scattered and clinical descriptions can be obscure The new Physician's Guide with the additional more than 600 diseases now featured documents 1200 conditions grouped according to type of disorder organ system affected e g liver kidney etc or phenotype e g neurological hepatic etc It includes relevant clinical findings and highlights the pathological values for diagnostic metabolites Guidance on appropriate biochemical genetic testing is also provided and established experimental therapeutic protocols are described with recommendations on follow up and monitoring The authors are acknowledged experts and the book is a valuable desk reference for all who deal with inherited metabolic diseases Chapter 73 is available open access under a Creative Commons Attribution 4.0 International License via link [springer.com](https://www.springer.com) Detection of Non-Amplified Genomic DNA Giuseppe Spoto, Roberto Corradini, 2012-07-06 This book offers an overview of state of the art in non amplified DNA detection methods and provides chemists biochemists biotechnologists and material scientists with an introduction to these methods In fact all these fields have dedicated resources to the problem of nucleic acid detection each contributing with their own specific methods and concepts This book will explain the basic principles of the different non amplified DNA detection methods available highlighting their respective advantages and limitations Non amplified DNA detection can be achieved by adopting different techniques Such techniques have allowed the commercialization of innovative platforms for DNA detection that are expected to break into the DNA diagnostics market The enhanced sensitivity required for the detection of non amplified genomic DNA has prompted new strategies that can achieve ultrasensitivity by combining specific materials with specific detection tools Advanced materials play multiple roles in ultrasensitive detection Optical and electrochemical detection tools are among the most widely investigated to analyze non amplified nucleic acids

Biosensors based on piezoelectric crystal have been also used to detect unamplified genomic DNA The main scientific topics related to DNA diagnostics are discussed by an outstanding set of authors with proven experience in this field

**Literature Search** National Library of Medicine (U.S.),1984 *Pediatric Urology* Warren T. Snodgrass,2013-05-13 Pediatric Urology Evidence for Optimal Patient Management provides pediatric urologists the information needed for state of the art patient care Chapters are organized around pertinent clinical questions within major areas of pediatric urology answered using the best available data while also reporting areas for which there is poor evidence The text includes randomized controlled trials and prospective observational studies tables that summarize important studies and figures that illustrate algorithms with best options for management and their expected results With an easy to use format not found in other volumes Pediatric Urology Evidence for Optimal Patient Management is an indispensable and unique resource for experienced pediatric urologists pediatric surgeons general urologists with an interest in pediatric urology as well as fellows and residents in training

**Diagnostic Ultrasound in Pregnancy** Charlotte Kenton,1984 **Textbook of Pediatric Gastroenterology, Hepatology and Nutrition** Stefano Guandalini,Anil Dhawan,2021-11-24 The latest edition of this textbook provides a comprehensive state of the art overview of the major issues specific to the field of pediatric gastroenterology hepatology and nutrition The textbook begins with a section on gastroenterology and nutrition that presents the overall scope of issues encountered in children suffering from disorders of the gastrointestinal tract pancreas and or presenting nutritional issues as well as current and future prospects on the use of prebiotics probiotics and postbiotics The second section is centered around hepatology reviewing congenital and acquired disorders of the biliary tract and liver as well as analyzing available diagnostic and therapeutic procedures and future perspectives Written by experts in the field Textbook of Pediatric Gastroenterology Hepatology and Nutrition A Comprehensive Guide Second Edition is an indispensable resource for students trainees and clinicians sure to distinguish itself as the definitive reference on this topic

*Diagnostic Ultrasound E-Book* Carol M. Rumack,Deborah Levine,2023-10-06 Spanning a wide range of medical specialties and practice settings Diagnostic Ultrasound 6th Edition provides complete detailed information on the latest techniques for ultrasound imaging of the whole body image guided procedures fetal obstetric and pediatric imaging and much more This thoroughly revised two volume set edited by Drs Carol M Rumack and Deborah Levine remains the most comprehensive and authoritative ultrasound resource available Up to date guidance from experts in the field keep you abreast of expanding applications of this versatile imaging modality and help you understand the how and why of ultrasound use and interpretation Covers all aspects of diagnostic ultrasound with sections for Physics Abdominal Pelvic Small Parts Vascular Obstetric and Pediatric Sonography Contains 5 000 images throughout including 2D and 3D imaging as well as the use of contrast agents and elastography Includes a new section on setting up a contrast lab for clinical practice and a new chapter on hemodialysis Features new coverage of the parotid salivary and submandibular glands as well as the retroperitoneum which now includes a section on endoleaks with

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therapies in cancer patients Identification of cell free circulating epigenomic biomarkers for early diagnosis and response to therapies in breast cancer patients Expanding frontiers in liquid biopsy discovery and validation of circulating biomarkers in renal cell carcinoma and bladder cancer and much more This comprehensive work explores the various types of biomarkers their significance in early cancer detection prognostication and evaluation of therapeutic responses It covers cutting edge research on circulating tumor cells cell free DNA exosomes and other biomarkers that can revolutionize cancer management Additionally it discusses the challenges advancements and future prospects of utilizing circulating biomarkers in personalized cancer care Provides a comprehensive overview of circulating biomarkers used in cancer diagnosis prognosis and treatment response monitoring Discusses the clinical utility of circulating biomarkers in early cancer detection prognostication treatment selection and monitoring therapeutic responses Explores the latest technologies and methodologies for the detection quantification and analysis of circulating biomarkers in cancer patients **Inborn**

**Metabolic Diseases** Jean-Marie Saudubray, Matthias R. Baumgartner, John H. Walter, 2016-11-10 This work is recognised as the standard textbook for professionals involved in the diagnosis and management of inborn errors of metabolism IEM and an essential resource in this multidisciplinary field For the 6th edition all 43 chapters have been newly written or revised by authors with particular expertise in their subject areas Contents A clinical and biochemical approach to the recognition and diagnosis of IEM with algorithms to symptoms signs and syndromes in patients of all ages Emergency treatments Medications Separate comprehensive sections on IEM of Carbohydrates Mitochondrial Energy Amino and organic acids Vitamin responsive defects Neurotransmitter and Small peptides Lipid and Bile Acids Nucleic Acid and Heme Organelles Disorders affecting the synthesis and remodelling of complex lipids and fatty acid homeostasis are now included *Genetic Counseling* Geraldine D Nowak, 1978 **Creasy and Resnik's Maternal-Fetal Medicine: Principles and Practice**

**E-Book** Charles J. Lockwood, Thomas Moore, Joshua Copel, Robert M Silver, Robert Resnik, 2018-08-07 Long recognized as the authoritative leader in the field Creasy and Resnik's Maternal Fetal Medicine 8th Edition continues to provide the latest evidence based guidelines for obstetric and neonatal management helping you minimize complications and offer patients the best possible care Written by renowned experts in obstetrics gynecology and perinatology this comprehensive resource has been thoroughly updated and reflects new information in every area including recent tremendous advances in genetics imaging and more Focuses on complicated obstetric issues highlighting the most commonly encountered anomalies and providing clear guidelines for obstetric and neonatal management Offers comprehensive updates on rapidly changing topics including a completely revised section on genetics and genetic technology for prenatal diagnoses as well as an expanded imaging section on abdominal urogenital and skeletal imaging Includes four new chapters Molecular Genetic Technology MRI in Obstetrical Imaging Obesity in Pregnancy and Pregnancy as a Window to Future Health Features numerous flow charts for quick access to diagnosis and treatment protocols and to clarify complex material Presents the knowledge and

expertise of new editors Dr Joshua Copel an expert in the field of fetal therapy who has pioneered new diagnostic techniques for unborn patients and their mothers and Dr Robert Silver a leader in the maternal fetal medicine community

Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease Roger N. Rosenberg, Juan M.

Pascual, 2014-10-28 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease Fifth Edition provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to the majority of neurologic and psychiatric disease. A favorite of over three generations of students, clinicians, and scholars, this new edition retains and expands the informative, concise, and critical tone of the first edition. This is an essential reference for general medical practitioners, neurologists, psychiatrists, geneticists, and related professionals, and for the neuroscience and neurology research community. The content covers all aspects essential to the practice of neurogenetics to inform clinical diagnosis, treatment, and genetic counseling. Every chapter has been thoroughly revised or newly commissioned to reflect the latest scientific and medical advances by an international team of leading scientists and clinicians. The contents have been expanded to include disorders for which a genetic basis has been recently identified, together with abundant original illustrations that convey and clarify the key points of the text in an attractive, didactic format. Previous editions have established this book as the leading tutorial reference on neurogenetics. Researchers will find great value in the coverage of genomics, animal models, and diagnostic methods, along with a better understanding of the clinical implications. Clinicians will rely on the coverage of the basic science of neurogenetics and the methods for evaluating patients with biochemical abnormalities or gene mutations, including links to genetic testing for specific diseases. Comprehensive coverage of the neurogenetic foundation of neurological and psychiatric disease. Detailed introduction to both clinical and basic research implications of molecular and genetic understanding of the brain. Detailed coverage of genomics, animal models, and diagnostic methods, with new coverage of evaluating patients with biochemical abnormalities or gene mutations.

**Complement Therapeutics** John D. Lambris, V. Michael Holers, Daniel Ricklin, 2012-09-19 This book highlights progress and trends in the rapidly evolving field of complement-related drug discovery and spotlights examples of clinical applications. As an integral part of innate immunity and critical mediator in homeostatic and inflammatory processes, the human complement system has been identified as a contributor to a large number of disorders, including ocular, cardiovascular, metabolic, autoimmune, and inflammatory diseases, as well as in ischemia/reperfusion injury, cancer, and sepsis. In addition, complement is often involved in adverse immune reactions to biomaterials, cell and organ transplants, or drug delivery systems. Although the complement cascade, with its close to 50 extracellular protein targets, has long been recognized as an attractive system for therapeutic modulation, the past few years have seen a particularly strong boost in interest. Fueled by novel research insight and the marketing of the first complement-targeted drugs, a plethora of highly creative treatment approaches and potent drug candidates have recently emerged and are currently evaluated in disease models and clinical

trials The chapters in this book cover a wide range of topics related to the development of complement therapeutics ranging from the molecular and functional description of complement targets to the presentation of novel inhibitors improved treatment strategies as well as examples of disease models and clinical applications The broad and up to date overview on a highly versatile and dynamic field renders this book an indispensable source of information for researchers and clinicians dealing with therapeutic and disease related aspects of the human complement system **Donald School Basic Textbook of Ultrasound in Obstetrics & Gynecology** V. D'Addario, 2014-11-30 The second edition of this book has been fully updated to provide clinicians with developments in the role of ultrasound in the care of pregnant women Beginning with an overview of ultrasound techniques the following chapters discuss normal foetal anatomy and abnormal signs providing in depth coverage of the appearance of malformations in different systems of the foetus anatomy including gastrointestinal urinary thorax skeletal and the central nervous system The final sections examine ultrasound of the placenta uterus fallopian tubes the umbilical cord ovaries and much more Separate chapters are dedicated to twins and evaluation of benign pelvic masses Written by an experienced specialist from Bari University Medical School Italy this new edition has been fully revised and includes new information on recent techniques such as 3D and 4D ultrasound More than 1000 ultrasound images and illustrations enhance the comprehensive text Key points Second edition presenting clinicians with recent developments in obstetric and gynaecological ultrasound Includes more than 1000 ultrasound images and illustrations Written by expert from Bari University Medical School Italy Previous edition published in 2008 *Molecular Pathology in Clinical Practice* Debra G.B. Leonard, 2016-02-02 This authoritative textbook offers in depth coverage of all aspects of molecular pathology practice and embodies the current standard in molecular testing Since the successful first edition new sections have been added on pharmacogenetics and genomics while other sections have been revised and updated to reflect the rapid advances in the field The result is a superb reference that encompasses molecular biology basics genetics inherited cancers solid tumors neoplastic hematopathology infectious diseases identity testing HLA typing laboratory management genomics and proteomics Throughout the text emphasis is placed on the molecular variations being detected the clinical usefulness of the tests and important clinical and laboratory issues The second edition of *Molecular Pathology in Clinical Practice* will be an invaluable source of information for all practicing molecular pathologists and will also be of utility for other pathologists clinical colleagues and trainees **Genetic Disorders and the Fetus** Aubrey Milunsky, Jeff M. Milunsky, 2011-09-07 Highly Commended in the Obstetrics and Gynaecology category of the 2010 BMA Medical Book Competition Brand new edition of the world's leading text on prenatal diagnosis This 6th Edition of *Genetic Disorders and the Fetus* maintains its pre eminence as the major repository of facts about prenatal diagnosis It provides a critical analysis and synthesis of established and new knowledge based on the long experience of authorities in their respective fields A broad international perspective is presented through authoritative contributions from authors in 11 countries All chapters and guidelines have been updated to



reflect contemporary practice New chapters have been introduced on The use of chromosomal microarrays in prenatal diagnosis The social legal and public policy issues with special reference to international approaches The important peroxisomal and related fatty acid oxidation disorders Extensive tables and clear illustrations assist in differential diagnosis gene identification and diagnostic modes The recognition of many new and unresolved challenges should provide inspiration for novel research initiatives The guidance provided and the insights and perspectives of these authors make this volume a valuable and indispensable resource for all whose focus is securing fetal health through prenatal diagnosis Genetic Disorders and the Fetus Diagnosis Prevention and Treatment is an essential resource for all engaged in prenatal genetic diagnosis especially obstetricians maternal fetal medicine specialists medical geneticists genetic counsellors and pediatricians but also many other specialties

*Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease, Seventh Edition* Roger N. Rosenberg, Juan M. Pascual, 2024-10-04 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease Seventh Edition provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to neurologic and psychiatric disease This volume has been thoroughly revised and includes newly commissioned chapters on ethics genetic counselling and gene therapy for the central nervous system disorders A favorite of over four generations of students clinicians and scholars this new edition retains and expands the informative concise and critical tone of the previous edition This is an essential reference for general medical practitioners neurologists psychiatrists geneticists and related professionals and for the neuroscience and neurology research community at large Both volumes combined provide a comprehensive coverage on the neurogenetic foundation of neurological and psychiatric disease This volume provides a detailed introduction on both the clinical and basic research implications of molecular and genetics surrounding the brain Includes new chapters on genomics of human neurological disorders CRISPR and genome engineering

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