

中华生殖与避孕杂志[®]

原刊名《生殖与避孕》

CHINESE JOURNAL OF REPRODUCTION AND CONTRACEPTION

月刊 1980年12月创刊 第45卷 第8期 2025年8月25日出版



官方微信

主 管

中国科学技术协会

主 办

中华医学会
上海市生物医药技术研究院
复旦大学附属妇产科医院

编 辑

中华生殖与避孕杂志
编辑委员会
200237,上海市老沪闵路779号
电话:(021)64438169
传真:(021)64438975
Email:rande@sibpt.cn
http://zhshzybyzz.yiigle.com

总编辑

乔 杰

编辑部主任

王 健

出 版

《中华医学杂志》社有限责任公司
100710,北京市东西大街42号
电话(传真):(010)51322059
Email:office@cmaph.org

印 刷

上海界龙中报印务有限公司

发 行

范围:公开
国内:中国邮政集团公司
上海分公司
国外:中国国际图书贸易集团
责任有限公司
(北京399信箱,100048)
代号 BM 389

订 购

全国各地邮政局
邮发代号4-928

邮 购

中华生殖与避孕杂志编辑部
200237,上海市老沪闵路779号
电话:(021)64438169,64438975
Email:rande@sibpt.cn

定 价

每期35.00元,全年420.00元

中国标准连续出版物号

ISSN 2096-2916
CN 10-1441/R

2025年版权归主办者所有

未经授权,不得转载、摘编本刊
文章,不得使用本刊的版式设计

除非特别声明,本刊刊出的所有
文章不代表中华医学会和本刊
编委会的观点

本刊如有印装质量问题,请向本刊
编辑部调换

目 次

生殖遗传专栏

- Y染色体微缺失遗传筛查专家建议(2025年) 757
中国医师协会生殖医学专业委员会 中国人体健康科技促进会
男科学专业委员会
辅助生殖人群扩展性携带者筛查的临床应用及挑战 763
林丽晶 徐晨明
空卵泡综合征的病因与治疗 769
陈晓蕾 杨萍 王奕锐 陈子江 赵涵
嵌合型胚胎移植的研究进展 775
刘家柳 徐艳文

临床研究

- 同型半胱氨酸与反复妊娠丢失的相关性及其对反复妊娠丢失
患者IVF/ICSI-ET妊娠结局的影响 779
刘曼曼 张瑞 张贺博 原梦帆 任炳楠 张俊韦 李枫
张文娟 刘朝朝 管一春
女性体质量指数对供精人工授精生育结局的影响 787
张清健 祝晓丽 詹泽虎 蔡晓林 李艳 李秋华
拮抗剂方案新鲜胚胎移植后单用地屈孕酮片与联合用阴道
黄体酮凝胶的临床结局比较 794
黄国斐 丁书芳 林苏霞 罗龙丹 叶天民
优质胚胎冷冻保存时长对临床妊娠结局影响的回顾性
队列研究 802
郭小妮 刘晓洁 张孝东 黄国宁 张琦

实验研究

- 脐带血单个核细胞宫腔灌注对薄型子宫内膜建模小鼠
子宫内膜厚度及容受性的影响 809
刘超 程兰兰 谭丽

循证医学

- 体外受精促排卵领域指南与共识的现况分析与质量评价 818
高媛 宋再伟 姜丹 杨蕊 黄育文 李蓉 赵荣生
CYP17A1基因突变所致先天性肾上腺皮质增生症合并
不孕症患者的最佳助孕治疗策略:一项系统综述 826
侯志金 刘洋 杨晓玲 李东娅 孟昱时

个案报道

基于二代测序的PGT技术偶然发现隐匿性复杂染色体重排男性携带者
1例报道 833
邓杰 马宁 周多 王火鸟 周俏苗

综 述

人工智能在子宫腺肌病精准化诊疗中的研究进展 837
余璐萍 梅洁
中性粒细胞在子宫内膜异位症中的研究进展 841
曹芸蕾 杜晶
无创胚胎植入前非整倍体检测技术的研究进展及展望 846
黄澳森 张印峰 刘聪 韩莹 罗海宁
丙戊酸对癫痫男性生殖功能影响的研究进展 851
李柯婧 李沛 尹争 李西军 李欣
哺乳动物原核形成和消失的分子机制研究进展 856
王超莹 姚桂东

经验交流

人类精子库供精者遗传咨询的实践经验 861
刘浏 朱勇 梁国庆 朱宏 江峰

《中华生殖与避孕杂志》第三届编辑委员会名单 768
《中华生殖与避孕杂志》第三届通讯编辑委员会名单 793
《中华生殖与避孕杂志》稿约 封三
本期责任编委 黄荷凤 徐艳文 本期责任编辑 宋培培 本期责任排版 年丽红

CHINESE JOURNAL OF REPRODUCTION AND CONTRACEPTION

(Original title: *Reproduction and Contraception*)

Monthly

Established in December 1980

Volume 45, Number 8

August 25, 2025



Responsible Institution

China Association for Science and Technology

Sponsor

Chinese Medical Association,
Shanghai Institute for Biomedical and Pharmaceutical Technologies,
Obstetrics and Gynecology Hospital of Fudan University

Editing

Editorial Board of Chinese Journal of Reproduction and Contraception
779 Laohumin Road, Shanghai 200237, China
Tel: 0086-21-64438169
Fax: 0086-21-64438975
Email: rande@sibpt.cn
<http://zhshzybyzz.yiigle.com>

Editor-in-Chief

Qiao Jie

Managing Director

Wang Jian

Publishing

Chinese Medical Journals
Publishing House Co., Ltd.
42 Dongsi Xidajie, Beijing 100710, China
Tel(Fax): 0086-10-51322059
Email: office@cmaph.org

Printing

Shanghai Jielong Zhongbao Printing Co., Ltd

Overseas Distributor

China International Book Trading Corporation
P.O. Box 399, Beijing 100048, China
Code No.M389

Mail-Order

Editorial Board of Chinese Journal of Reproduction and Contraception
779 Laohumin Road, Shanghai 200237, China
Tel: 0086-21-64438169
Fax: 0086-21-64438975
Email: rande@sibpt.cn

CSSN

ISSN 2096-2916
CN 10-1441/R

Copyright © 2025 by the all sponsors

No content published by the journals of Chinese Medical Association may be reproduced or abridged without authorization. Please do not use or copy the layout and design of the journals without permission.

All articles published represent the opinions of the authors, and do not reflect the official policy of the Chinese Medical Association or the Editorial Board, unless this is clearly specified.

CONTENTS IN BRIEF

Reproductive Genetics Column

- Experts advice of Chinese experts on genetic screening for Y chromosome microdeletions (2025)** 757
Reproductive Medicine Professional Committee of Chinese Medical Doctor Association, Andrology Professional Committee of China Association for Promotion of Health Science and Technology
- Expanded carrier screening in assisted reproduction: current clinical applications and emerging challenges** 763
Lin Lijing, Xu Chenming
- Etiology and treatment of empty follicle syndrome** 769
Chen Xiaolei, Yang Ping, Wang Yikun, Chen Zijiang, Zhao Han
- Research progress of mosaic embryo transfers** 775
Liu Jialiu, Xu Yanwen

Clinical Studies

- Association between homocysteine and recurrent pregnancy loss (RPL) and its effects on IVF/ICSI-ET outcomes in RPL patients** 779
Liu Manman, Zhang Rui, Zhang Hebo, Yuan Mengfan, Ren Bingnan, Zhang Junwei, Li Feng, Zhang Wenjuan, Liu Chaozhao, Guan Yichun
- Effect of female body mass index on fertility outcomes of artificial insemination with donor sperm** 787
Zhang Qingjian, Zhu Xiaoli, Zhan Zehu, Cai Xiaolin, Li Yan, Li Qiuhua
- Comparison of clinical outcomes between oral dydrogesterone and vaginal progesterone gel plus oral dydrogesterone after fresh embryo transfer with GnRH antagonist protocol** 794
Huang Yuanfei, Ding Shufang, Lin Suxia, Luo Longdan, Ye Tianmin
- Impact of cryostorage duration on clinical outcomes: a retrospective cohort study of vitrified high-quality embryos** 802
Guo Xiaoni, Liu Xiaojie, Zhang Xiaodong, Huang Guoning, Zhang Qi

Laboratory Study

- Impact of umbilical cord blood mononuclear cells intrauterine perfusion on endometrial thickness and endometrial receptivity in mice with thin endometrium** 809
Liu Chao, Cheng Lanlan, Tan Li

Evidence-based Medicine

- Current situation analysis and quality evaluation of guidelines and consensus in ovarian stimulation for *in vitro* fertilization** 818
Gao Yuan, Song Zaiwei, Jiang Dan, Yang Rui, Huang Yuwen, Li Rong, Zhao Rongsheng
- Optimal assisted reproductive strategies for infertile patients with congenital adrenal hyperplasia associated with CYP17A1 gene mutations: a systematic review** 826
Hou Hijin, Liu Yang, Yang Xiaoling, Li Dongya, Meng Yushi

Case Report

Incidental identification of a cryptic complex chromosomal rearrangement in a male carrier by next-generation sequencing-based PGT: a case report	833
<i>Deng Jie, Ma Ning, Zhou Duo, Wang Huoniao, Zhou Qiaomiao</i>	

Reviews

Research progress of artificial intelligence in the precision diagnosis and treatment of adenomyosis	837
<i>Yu Luping, Mei Jie</i>	
Research progress of neutrophils in endometriosis	841
<i>Cao Yunlei, Du Jing</i>	
Research progress and prospects of non-invasive preimplantation genetic testing technology	846
<i>Huang Aomiao, Zhang Yinfeng, Liu Cong, Han Ying, Luo Haining</i>	
Progress in study of effect of valproic acid on male reproductive function in epileptic adults	851
<i>Li Kejing, Li Pei, Yin Zheng, Li Xijun, Li Xin</i>	
Research advances in the molecular mechanisms underlying pronuclear formation and disappearance in mammals	856
<i>Wang Chaoying, Yao Guidong</i>	

Experience Exchange

Practical experience in genetic counseling for sperm donors in human sperm bank	861
<i>Liu Liu, Zhu Yong, Liang Guoqing, Zhu Hong, Jiang Feng</i>	

DOI: 10.3760/cma.j.cn101441-20250122-00033

收稿日期 2025-01-22 本文编辑 宋培培

引用本文: 中国医师协会生殖医学专业委员会, 中国人体健康科技促进会男科学专业委员会. Y 染色体微缺失遗传筛查专家建议 (2025 年)[J]. 中华生殖与避孕杂志, 2025, 45(8): 757-762. DOI: 10.3760/cma.j.cn101441-20250122-00033.

Y 染色体微缺失遗传筛查专家建议 (2025 年)

中国医师协会生殖医学专业委员会 中国人体健康科技促进会男科学专业委员会

通信作者: 李铮, 上海交通大学医学院附属第一人民医院泌尿中心

男科, 上海 200025, Email: lizhengboshi@sjtu.edu.cn; 张锋,

复旦大学医学遗传研究院 宋庆龄妇幼健康研究院, 上海 200032,

Email: zhangfeng@fudan.edu.cn; 熊承良, 华中科技大学同济医学

院, 武汉 430030, Email: clxiong951@sina.com

【摘要】 在精子发生障碍病因学诊断中, Y 染色体微缺失遗传筛查至关重要。2023 年欧洲男科学学会/欧洲分子遗传学质量网络对 Y 染色体筛查指南进行修订, 2023 年我国已经发布《Y 染色体微缺失检测试剂盒》行业标准 (YY/T 1893-2023), Y 染色体微缺失筛查日趋规范化。依据中国 YqAZF 微缺失筛查室间质量评价的数据, 参与质控的实验室分子诊断错误率大幅下降, 诊断报告质量也显著改善。但诸多医疗机构在开展筛查时, 仍然选择不同的筛查位点, 筛查报告也缺乏统一规范。本专家建议旨在更新 Y 染色体微缺失遗传筛查标准、筛查建议, 规范筛查报告的模板, 为从事生殖健康领域的相关人员提供专家咨询和诊疗建议。

【关键词】 男性不育; 基因检测; Y 染色体微缺失; 精子发生障碍

基金项目: 国家重点研发计划 (2022YFC2702700); 国家自然科学基金 (82371607、82271638、82171586)

实践指南注册: 国际实践指南注册与透明化平台, PREPARE-2024CN1231

Experts advice of Chinese experts on genetic screening for Y chromosome microdeletions (2025)

*Reproductive Medicine Professional Committee of Chinese Medical Doctor Association;
Andrology Professional Committee of China Association for Promotion of Health
Science and Technology*

*Corresponding authors: Li Zheng, Department of Andrology, Urology Center, the First
People's Hospital Affiliated with Shanghai Jiao Tong University School of Medicine,
Shanghai 200025, China, Email: lizhengboshi@sjtu.edu.cn; Zhang Feng, Institute of
Medical Genetics and Genomics, Fudan University; Soong Ching Ling Institute of
Maternity and Child Health, Shanghai 200032, China, Email: zhangfeng@fudan.edu.
cn; Xiong Chengliang, Tongji Medical College, Huazhong University of Science and
Technology, Wuhan 430030, China, Email: clxiong951@sina.com*

【Abstract】 Genetic screening for Y chromosome microdeletions is crucial in the etiological diagnosis of spermatogenic dysfunction. In 2023, the European Academy of Andrology/European Molecular Genetics Quality Network updated the guidelines on Y chromosome microdeletions screening. In recent years, China had already released guidelines and industry standards on test kits for genetic screening for Y chromosome microdeletions (YY/T 1893-2023). According to data from China Association on YqAZF Microdeletion External Quality Assessment, the error rates significantly decreased in molecular diagnostics among participating laboratories, and the quality of diagnostic reports had improved. But many medical institutions incorporated different genetic screening loci in their protocols and the screening reports also lack unified standards. Therefore, updating the domestic consensus was particularly necessary. This experts advice aimed to update the standards and recommendations for genetic screening for Y chromosome microdeletions and to standardize screening report template, providing expert consultation and diagnostic recommendations for professionals in the reproductive health field.

【 Key words 】 Infertility, male; Genetic testing; Y chromosome microdeletion; Spermatogenic dysfunction

Fund program: National Key R&D Program of China (2022YFC2702700); National Natural Science Foundation of China (82371607, 82271638, 82171586)

Practice guideline registration: Practice Guideline REgistration for transPAREncy, PREPARE-2024CN1231

·生殖遗传专栏·

DOI: 10.3760/cma.j.cn101441-20250411-00185

收稿日期 2025-04-11 本文编辑 宋培培

引用本文: 林丽晶, 徐晨明. 辅助生殖人群扩展性携带者筛查的临床应用及挑战[J]. 中华生殖与避孕杂志, 2025, 45(8): 763-768. DOI: 10.3760/cma.j.cn101441-20250411-00185.

辅助生殖人群扩展性携带者筛查的临床应用及挑战

林丽晶 徐晨明

复旦大学附属妇产科医院遗传中心, 上海 200001

通信作者: 徐晨明, Email: chenming_xu2006@163.com

【摘要】 扩展性携带者筛查是一种新型遗传风险评估工具, 通过高通量测序技术可一次性检测数百种常染色体隐性或 X 连锁隐性单基因遗传病的携带风险, 为辅助生殖人群提供生育的遗传风险分层和决策支持。然而, 扩展性携带者筛查仍面临疾病纳入标准不统一、变异解读复杂等挑战。未来需基于中国人群遗传数据开发适合中国人群的携带者筛查产品, 完善数据安全和隐私保护政策, 建立培训体系提升生殖遗传咨询能力, 以推动辅助生殖领域扩展性携带者筛查的精准应用。

【关键词】 生殖技术, 辅助; 扩展性携带者筛查; 二代测序; 携带者筛查; 植入前遗传学检测

基金项目: 国家重点研发计划 (2023YFC2705600、2023YFC2705601); 上海市科委科技计划项目 (22S31901500)

Expanded carrier screening in assisted reproduction: current clinical applications and emerging challenges

Lin Lijing, Xu Chenming

Genetics Center of Obstetrics and Gynecology, Obstetrics and Gynecology Hospital, Fudan University, Shanghai 200001, China

Corresponding author: Xu Chenming, Email: chenming_xu2006@163.com

【 Abstract 】 Expanded carrier screening (ECS) is a novel genetic risk assessment approach that leverages next-generation sequencing technologies to simultaneously identify carrier status for hundreds of autosomal recessive or X-linked recessive monogenic disorders in a single assay. This strategy enables stratified risk evaluation and supports informed reproductive decision-making, particularly among individuals undergoing assisted reproductive technologies (ARTs). Despite its increasing utility, the clinical implementation of ECS remains constrained by several challenges, including inconsistencies in screening panel inclusion criteria and the complexity of genetic variant interpretation. To promote the precision application of ECS in ARTs populations, future efforts should prioritize the development of population-specific panels informed by comprehensive genetic data from Chinese cohorts. In addition, strengthening data security and privacy protection policies, along with establishing specialized training programs in reproductive genetic counseling, will be essential for ensuring the responsible and effective integration of ECS into clinical practice.

【Key words】 Reproductive techniques, assisted; Expanded carrier screening; Next-generation sequencing; Carrier screening; Preimplantation genetic testing

Fund program: National Key Research and Development Program of China (2023YFC2705600, 2023YFC2705601); Shanghai Municipal Commission of Science and Technology Program (22S31901500)

生殖遗传专栏

DOI: 10.3760/cma.j.cn101441-20250330-00160

收稿日期 2025-03-30 本文编辑 孙敏

引用本文: 陈晓蕾, 杨萍, 王奕锟, 等. 空卵泡综合征的病因与治疗[J]. 中华生殖与避孕杂志, 2025, 45(8): 769-774. DOI: 10.3760/cma.j.cn101441-20250330-00160.

空卵泡综合征的病因与治疗

陈晓蕾¹ 杨萍² 王奕锟¹ 陈子江¹ 赵涵¹

¹ 山东大学附属生殖医院生殖科 山东大学生殖医学与子代健康全

国重点实验室, 济南 250012; ² 山东第一医科大学附属省立医院

妇产科, 济南 250001

通信作者: 赵涵, Email: hanzh80@sdu.edu.cn

【摘要】 空卵泡综合征(empty follicle syndrome, EFS)是指在控制性卵巢刺激过程中卵泡发育正常,但无法获得卵母细胞的情况,是一种病因复杂的疾病,分为真性EFS和假性EFS。真性EFS病因复杂,与遗传因素和卵巢功能有关, *LHCGR* 和 *ZP* 基因突变是真性EFS的主要遗传病因,但大部分患者原因不明。真性EFS强调对因治疗,各种方法治疗无效后可能需要选择供卵治疗。假性EFS病因基本明确,与触发排卵药物的不当使用有关,可以通过二次注射人绒毛膜促性腺激素或更换用药方案等进行治疗。本文从真性及假性EFS的病因和精准治疗两个方面进行综述,总结了近年来EFS的最新诊治进展,旨在为临床诊治和进一步病因研究提供方向。

【关键词】 生殖技术, 辅助; 空卵泡综合征; *LHCGR* 突变; *ZP* 基因突变

基金项目: 国家自然科学基金重大项目(82192874); 国家自然科学基金创新研究群体项目(82421004)

Etiology and treatment of empty follicle syndrome

Chen Xiaolei¹, Yang Ping², Wang Yikun¹, Chen Zijiang¹, Zhao Han¹

¹ Department of Reproductive Medicine, Center for Reproductive Medicine, State Key Laboratory of Reproductive Medicine and Offspring Health, Shandong University, Jinan 250012, China; ² Department of Obstetrics and Gynecology, Shandong Provincial Hospital Affiliated to Shandong First Medical University, Jinan 250001, China

Corresponding author: Zhao Han, Email: hanzh80@sdu.edu.cn

【 Abstract 】 Empty follicle syndrome (EFS), defined by the failure to retrieve oocytes despite normal follicle development following controlled ovarian stimulation, is a multifactorial condition in assisted reproductive treatment and categorized into genuine EFS (GEFS) and false EFS (FEFS). The etiology of GEFS is complex, involving genetic factors and ovarian dysfunction, with mutations in *LHCGR* and *ZP* genes being the major causes. However, the underlying cause remains unknown in most patients. Targeted therapies based on specific etiologies are crucial in GEFS management. Oocyte donation is considered as a final viable option when other precision treatments prove ineffective. The etiology of FEFS is relatively clear, primarily associated with inappropriate administration of ovulation-triggering drugs, which can be rescued by a second injection of human chorionic gonadotropin or change the medication regimen. This review summarizes current advances in the diagnosis and treatment of EFS, aiming to provide guidance for clinical management and further etiology research.

【 Key words 】 Reproductive technology, assisted; Empty follicle syndrome; *LHCGR* mutations; *ZP* gene mutations

Fund program: Major Program of the National Natural Science Foundation of China (82192874); Science Fund for Creative Research Groups of the National Natural Science Foundation of China (82421004)

生殖遗传专栏

DOI: 10.3760/cma.j.cn101441-20250418-00192

收稿日期 2025-04-18 本文编辑 宋培培

引用本文: 刘家柳, 徐艳文. 嵌合型胚胎移植的研究进展[J]. 中华生殖与避孕杂志, 2025, 45(8): 775-778.

DOI: 10.3760/cma.j.cn101441-20250418-00192.

嵌合型胚胎移植的研究进展

刘家柳 徐艳文

中山大学附属第一医院生殖医学中心 广东省妇产科疾病临床医学

研究中心 广东省生殖医学重点实验室, 广州 510080

通信作者：徐艳文，Email: xuyanwen@mail.sysu.edu.cn

【摘要】 胚胎植入前遗传学检测 (preimplantation genetic testing, PGT) 技术的临床应用提示了染色体嵌合在人类植入前胚胎中普遍存在的现象。嵌合型胚胎移植后有机会获得健康活产，但其妊娠结局及子代安全性仍存在不确定性风险。本文结合最新研究进展，从嵌合型胚胎检出率、自我纠正机制、移植后的妊娠结局，以及子代安全性几个方面进行阐述，为嵌合型胚胎的临床管理提供参考依据。

【关键词】 胚胎移植； 嵌合； 植入前遗传学检测

基金项目： 国家重点研发计划 (2023YFC2705503)； 国家自然科学基金 (82471884)； 广东省自然科学基金 (2025A1515010982)； 广州市临床重大项目 (2023P-ZD19)； 广州市科学技术项目 (2023A04J2178)

Research progress of mosaic embryo transfers

Liu Jialiu, Xu Yanwen

Reproductive Medicine Center, the First Affiliated Hospital of Sun Yat-sen University; Guangdong Provincial Clinical Research Center for Obstetrical and Gynecological Diseases; Key Laboratory for Reproductive Medicine of Guangdong Province, Guangzhou 510080, China

Corresponding author: Xu Yanwen, Email: xuyanwen@mail.sysu.edu.cn

【Abstract】 The clinical application of preimplantation genetic testing (PGT) has revealed a prevalent incidence of chromosomal mosaicism in human embryos. Although mosaic embryo transfer can yield healthy live births, the risk of pregnancy outcomes and offspring safety remains uncertain. This article comprehensively evaluates the clinical value of mosaic embryo transfer based on current evidence, focusing on the incidence of mosaicism, self-correction mechanisms, pregnancy outcomes, and offspring safety of mosaic embryo transfers. The findings offer evidence-based guidance for the clinical management of mosaic embryos.

【Key words】 Embryo transfer; Mosaicism; Preimplantation genetic testing

Fund program: National Key Research and Development Program of China (2023YFC2705503); National Natural Science Foundation of China (82471884); Natural Science Foundation of Guangdong Province (2025A1515010982); Key Clinical Technique of Guangzhou (2023P-ZD19); Guangzhou Science and Technology Project (2023A04J2178)

临床研究

DOI: 10.3760/cma.j.cn101441-20250306-00109

收稿日期 2025-03-06 本文编辑 孙敏

引用本文：刘曼曼，张瑞，张贺博，等. 同型半胱氨酸与反复妊娠丢失的相关性及其对反复妊娠丢失患者 IVF/ICSI-ET 妊娠结局的影响 [J]. 中华生殖与避孕杂志，2025，45(8): 779-786. DOI: 10.3760/cma.j.cn101441-20250306-00109.

同型半胱氨酸与反复妊娠丢失的相关性及其对反复妊娠丢失患者 IVF/ICSI-ET 妊娠结局的影响

刘曼曼 张瑞 张贺博 原梦帆 任炳楠 张俊韦 李枫 张文娟

刘朝朝 管一春

郑州大学第三附属医院生殖健康医院, 郑州 450052

通信作者: 刘曼曼, Email: huakailuolanlmm@126.com

【摘要】 目的 探讨同型半胱氨酸 (homocysteine, Hcy) 与反复妊娠丢失 (recurrent pregnancy loss, RPL) 的相关性, 及其对行体外受精/卵胞质内单精子注射-胚胎移植 (*in vitro* fertilization/intracytoplasmic sperm injection and embryo transfer, IVF/ICSI-ET) 治疗 RPL 患者临床妊娠结局的影响。**方法** 本研究为回顾性队列研究, 选取 2020 年 12 月至 2024 年 3 月于郑州大学第三附属医院生殖健康医院就诊, 行 IVF/ICSI-ET 患者的临床资料, 根据妊娠丢失史分为对照组 (既往从未发生妊娠丢失, $n=1\ 027$) 及 RPL 组 ($n=743$), 比较两组患者外周血 Hcy 水平。采用多因素 logistic 回归校正混杂因素的影响, 分析 Hcy 是否是影响 RPL 的相关因素。将 RPL 患者根据 Hcy 四分位数分为 4 个亚组, 分别为 Q_1 亚组 ($\text{Hcy} < 7.03\ \mu\text{mol/L}$)、 Q_2 亚组 ($7.03\ \mu\text{mol/L} \leq \text{Hcy} < 8.63\ \mu\text{mol/L}$)、 Q_3 亚组 ($8.63\ \mu\text{mol/L} \leq \text{Hcy} < 10.44\ \mu\text{mol/L}$)、 Q_4 亚组 ($\text{Hcy} \geq 10.44\ \mu\text{mol/L}$), 进一步分析不同 Hcy 水平对 RPL 患者行 IVF/ICSI-ET 后妊娠结局的影响。**结果** ①对照组与 RPL 组患者基线情况: 女方年龄、男方年龄、女方体质指数 (body mass index, BMI)、不孕年限、不孕因素、外周血 Hcy 水平, 两组间差异均有统计学意义 (均 $P < 0.05$)。②多因素 logistic 回归模型校正女方年龄、男方年龄、女方 BMI、不孕年限、不孕因素后, 结果显示, Hcy 水平异常升高是 RPL 的独立危险因素 ($\text{aOR}=1.366$, 95% CI : $1.298 \sim 1.438$, $P < 0.001$)。③4 个亚组 RPL 患者基线情况: Q_1 亚组、 Q_2 亚组、 Q_3 亚组、 Q_4 亚组患者的窦卵泡计数组间比较差异有统计学意义 [$17.00 (11.00, 24.00)$, $15.00 (10.00, 24.00)$, $14.00 (7.00, 22.25)$, $15.50 (8.00, 22.00)$, $P=0.043$], 其余基线情况比较差异均无统计学意义 (均 $P > 0.05$)。④4 个亚组 RPL 患者妊娠结局比较: Q_1 亚组、 Q_2 亚组、 Q_3 亚组和 Q_4 亚组患者的流产率分别为 18.18% (18/99)、30.61% (30/98)、33.70% (31/92)、35.96% (32/89), 活产率分别为 44.26% (81/183)、36.17% (68/188)、32.80% (61/186)、30.65% (57/186), 流产率和活产率组间比较差异均有统计学意义 ($P=0.033$, $P=0.036$)。其中, Q_3 亚组流产率、 Q_4 亚组流产率及活产率均显著高于 Q_1 亚组, 且差异均具有统计学意义 (均 $q < 0.05$)。然而, 4 个亚组患者临床妊娠率及早期流产率差异均无统计学意义 (均 $P > 0.05$)。多因素 logistic 回归校正混杂因素后, 以 Q_1 亚组为对照, 其余各组与其相比, 临床妊娠率差异均无统计学意义 (均 $P > 0.05$)。 Q_3 亚组早期流产率 ($\text{aOR}=2.184$, 95% CI :

1.077~4.426, $P=0.030$)、 Q_4 亚组早期流产率 ($aOR=2.290$, 95% CI : 1.116~4.697, $P=0.024$) 均显著高于 Q_1 亚组; Q_3 亚组流产率 ($aOR=2.207$, 95% CI : 1.125~4.330, $P=0.021$)、 Q_4 亚组流产率 ($aOR=2.377$, 95% CI : 1.209~4.674, $P=0.012$) 均显著高于 Q_1 亚组; Q_3 亚组活产率 ($aOR=0.615$, 95% CI : 0.401~0.944, $P=0.026$)、 Q_4 亚组活产率 ($aOR=0.560$, 95% CI : 0.364~0.863, $P=0.009$) 均显著低于 Q_1 亚组, 差异均具有统计学意义。结论 Hcy 水平异常升高是 IVF/ICSI-ET 患者发生 RPL 的高危因素, 并可能对 RPL 患者 IVF/ICSI-ET 妊娠结局产生不利影响。

【关键词】 生殖技术, 辅助; 妊娠结局; 同型半胱氨酸; 反复妊娠丢失

基金项目: 国家重点研发计划 (2021YFC2700602); 河南省医学科技攻关计划 (联合共建) 项目 (LHGJ20190369)

Association between homocysteine and recurrent pregnancy loss (RPL) and its effects on IVF/ICSI-ET outcomes in RPL patients

Liu Manman, Zhang Rui, Zhang Hebo, Yuan Mengfan, Ren Bingnan, Zhang Junwei, Li Feng, Zhang Wenjuan, Liu Chaozhao, Guan Yichun

Reproductive Health Hospital of the Third Affiliated Hospital of Zhengzhou University, Zhengzhou 450052, China

Corresponding author: Liu Manman, Email: huakailuolanlmm@126.com

【 Abstract 】 **Objective** To investigate the association between homocysteine (Hcy) and recurrent pregnancy loss (RPL), as well as its impact on clinical pregnancy outcomes in patients undergoing *in vitro* fertilization/intracytoplasmic sperm injection and embryo transfer (IVF/ICSI-ET). **Methods** This retrospective cohort study collected clinical data from patients undergoing IVF/ICSI-ET at the Reproductive Health Hospital of the Third Affiliated Hospital of Zhengzhou University between December 2020 and March 2024. Patients were divided into no history of pregnancy loss group (named control group, $n=1\ 027$) and RPL group ($n=743$) based on history of pregnancy loss. Peripheral blood Hcy levels were compared between the two groups. Multivariate logistic regression was performed to adjust for confounding factors and determine whether Hcy is an independent risk factor for RPL. RPL patients were divided into four subgroups based on Hcy quartiles, named Q_1 subgroup ($Hcy<7.03\ \mu\text{mol/L}$), Q_2 subgroup ($7.03\ \mu\text{mol/L}\leq Hcy<8.63\ \mu\text{mol/L}$), Q_3 subgroup ($8.63\ \mu\text{mol/L}\leq Hcy<10.44\ \mu\text{mol/L}$), and Q_4 subgroup ($Hcy\geq 10.44\ \mu\text{mol/L}$), to further analyze the impact of Hcy level on pregnancy outcomes after IVF/ICSI-ET in these patients. **Results** 1) Baseline characteristics between control and RPL groups: statistically significant differences were observed in female age, male age, female body mass index (BMI), duration of infertility, cause of infertility, and peripheral blood Hcy levels (all $P<0.05$). 2) After adjusting for female age, male age, female BMI, duration of infertility, and cause of infertility via multivariate logistic regression, elevated Hcy levels was identified as an independent risk factor for RPL ($aOR=1.366$, 95% CI : 1.298–1.438, $P<0.001$). 3) Baseline characteristics of the four RPL subgroups: antral follicle count (AFC) differed significantly among Q_1 , Q_2 , Q_3 and Q_4 subgroups [17.00 (11.00, 24.00), 15.00 (10.00, 24.00), 14.00 (7.00, 22.25),

15.50 [8.00, 22.00], $P=0.043$]. No statistically significant differences were observed in other baseline characteristics (all $P>0.05$). 4) Pregnancy outcomes across the four RPL subgroups: miscarriage rates in the Q_1 , Q_2 , Q_3 and Q_4 subgroups were 18.18% (18/99), 30.61% (30/98), 33.70% (31/92), and 35.96% (32/89), respectively, live birth rates were 44.26% (81/183), 36.17% (68/188), 32.80% (61/186), and 30.65% (57/186), respectively. Intergroup differences in miscarriage rate and live birth rate were statistically significant ($P=0.033$, $P=0.036$). Specifically, miscarriage rate in the Q_3 and Q_4 subgroups, and live birth rate in the Q_4 subgroup were significantly higher than those in the Q_1 subgroup (all $q<0.05$). However, no significant differences were observed in clinical pregnancy rate or early miscarriage rate among the four groups (all $P>0.05$). After adjusting for confounding factors using multivariate logistic regression, taking the Q_1 subgroup as the control, there were no statistically significant differences in the clinical pregnancy rate between the remaining groups and the Q_1 subgroup (all $P>0.05$). The early miscarriage rate in the Q_3 subgroup (aOR=2.184, 95% CI: 1.077–4.426, $P=0.030$) and the early miscarriage rate in the Q_4 subgroup (aOR=2.290, 95% CI: 1.116–4.697, $P=0.024$) were significantly higher than those in the Q_1 subgroup; the miscarriage rate in the Q_3 subgroup (aOR=2.207, 95% CI: 1.125–4.330, $P=0.021$) and the miscarriage rate in the Q_4 subgroup (aOR=2.377, 95% CI: 1.209–4.674, $P=0.012$) were significantly higher than those in the Q_1 subgroup; the live birth rate in the Q_3 subgroup (aOR=0.615, 95% CI: 0.401–0.944, $P=0.026$) and the live birth rate in the Q_4 subgroup (aOR=0.560, 95% CI: 0.364–0.863, $P=0.009$) were significantly lower than those in the Q_1 subgroup. **Conclusion** Elevated Hcy is a high-risk factor for RPL in IVF/ICSI-ET patients and may adversely affect pregnancy outcomes.

【Key words】 Reproductive technology, assisted; Pregnancy outcomes; Homocysteine; Recurrent pregnancy loss

Fund program: National Key Research and Development Program of China (2021YFC2700602); Henan Medical Science and Technology Key Research Plan (Joint Construction) Project (LHGJ20190369)

·临床研究·

DOI: 10.3760/cma.j.cn101441-20250221-00085

收稿日期 2025-02-21 本文编辑 王李艳

引用本文: 张清健, 祝晓丽, 詹泽虎, 等. 女性体质质量指数对供精人工授精生育结局的影响[J]. 中华生殖与避孕杂志, 2025, 45(8): 787-793. DOI: 10.3760/cma.j.cn101441-20250221-00085.

女性体质质量指数对供精人工授精生育结局的影响

张清健 祝晓丽 詹泽虎 蔡晓林 李艳 李秋华

国家卫生健康委员会男性生殖与遗传重点实验室 广东省生殖科学
研究所生殖中心, 广州 510600

通信作者: 李秋华, Email: 710971943@qq.com

【摘要】 目的 探索女性体质量指数 (body mass index, BMI) 对供精人工授精 (artificial insemination with donor sperm, AID) 妊娠结局的影响。**方法** 采用回顾性队列研究, 分析 2011 年 1 月至 2024 年 9 月在广东省生殖科学研究所生殖中心治疗的 4 484 对患者夫妇的 9 852 个 AID 治疗周期, 按照女性 BMI 分为 4 组, 低 BMI 组 ($BMI < 18.5 \text{ kg/m}^2$, $n=1\ 352$)、正常 BMI 组 ($18.5 \text{ kg/m}^2 \leq BMI < 24.0 \text{ kg/m}^2$, $n=6\ 548$)、超重组 ($24.0 \text{ kg/m}^2 \leq BMI < 28.0 \text{ kg/m}^2$, $n=1\ 538$) 和肥胖组 ($BMI \geq 28.0 \text{ kg/m}^2$, $n=414$)。比较各组间一般资料以及妊娠结局。采用生存分析 Kaplan-Meier 法计算 1~6 周期累积妊娠率。纳入年龄、基础卵泡刺激素、基础黄体生成素、子宫内膜厚度、临床诊断、治疗方案进行广义估计方程、单因素和多因素 logistic 和 Cox 回归分析, 探索女性 BMI 与临床妊娠率、自然流产率以及累积妊娠率相关性。**结果** ①低 BMI 组、正常 BMI 组、超重组和肥胖组的临床妊娠率和自然流产率差异均无统计学意义 (均 $P > 0.05$)。②AID 1~6 周期的累积妊娠率分别为 17.60%、31.60%、43.08%、54.37%、61.83%和 73.68%。③广义估计方程多因素分析显示, 女方年龄 ($OR=0.962$, 95% CI : 0.950~0.974, $P < 0.001$)、子宫内膜厚度 ($OR=1.040$, 95% CI : 1.011~1.069, $P=0.006$) 以及自然周期 ($OR=1.171$, 95% CI : 1.060~1.294, $P=0.002$) 影响临床妊娠率。与正常 BMI 组比较, 低 BMI 组、超重组和肥胖组临床妊娠率的组间差异均无统计学意义 (均 $P > 0.05$) ; 多因素 logistic 回归分析显示, 女方年龄 ($OR=1.051$, 95% CI : 1.012~1.091, $P=0.010$)、子宫内膜厚度 ($OR=0.920$, 95% CI : 0.847~1.000, $P=0.049$) 和多囊卵巢综合征 ($OR=1.927$, 95% CI : 1.044~3.556, $P=0.036$) 影响自然流产率; 与正常 BMI 组比较, 低 BMI 组、超重组和肥胖组自然流产率的组间差异均无统计学意义 (均 $P > 0.05$)。④Cox 回归分析显示, 女方年龄 ($HR=0.939$, 95% CI : 0.928~0.950, $P < 0.001$)、子宫内膜厚度 ($HR=1.039$, 95% CI : 1.013~1.066, $P=0.003$) 以及自然周期 ($HR=1.957$, 95% CI : 1.785~2.146, $P < 0.001$) 影响累积妊娠率。与正常 BMI 组比较, 低 BMI 组、超重组和肥胖组累积妊娠率的组间差异均无统计学意义 (均 $P > 0.05$)。**结论** 女性 BMI 的高低对于 AID 的临床妊娠率、自然流产率和累积妊娠率无显著影响。

【关键词】 供精人工授精; 临床妊娠率; 自然流产率; 累积妊娠率; 广义估计方程

Effect of female body mass index on fertility outcomes of artificial insemination with donor sperm

Zhang Qingjian, Zhu Xiaoli, Zhan Zehu, Cai Xiaolin, Li Yan, Li Qiuhua

*Key Laboratory of Male Reproduction and Genetics of National Health Commission;
Reproductive Center of Guangdong Institute of Reproductive Science, Guangzhou
510600, China*

Corresponding author: Li Qiuhua, Email: 710971943@qq.com

【Abstract】 Objective To explore the impact of female body mass index (BMI) on pregnancy outcomes in artificial insemination with donor sperm (AID). **Methods** A retrospective cohort study was conducted on 4 484 couples with 9 852 AID treatment cycles treated at Reproductive Center of Guangdong Institute of Reproductive Science from January 2011 to September 2024. Participants were divided into four groups based on BMI: low BMI group ($\text{BMI} < 18.5 \text{ kg/m}^2$), normal BMI group ($18.5 \text{ kg/m}^2 \leq \text{BMI} < 24.0 \text{ kg/m}^2$), overweight group ($24.0 \text{ kg/m}^2 \leq \text{BMI} < 28.0 \text{ kg/m}^2$), and obese group ($\text{BMI} \geq 28.0 \text{ kg/m}^2$). General characteristics and pregnancy outcomes were compared across groups. Kaplan-Meier survival analysis was used to calculate cumulative pregnancy rates from one to six cycles. Generalized estimating equations (GEE), univariate and multivariate logistic and Cox regression analysis were performed, adjusting for age, basal follicle-stimulating hormone, basal luteinizing hormone, endometrial thickness, clinical diagnosis, and treatment protocol, to explore correlations between female BMI and clinical pregnancy rate, spontaneous abortion rate, and cumulative pregnancy rate. **Results** 1) There were no statistically significant differences in clinical pregnancy rate and spontaneous abortion rate among the low BMI group, the normal BMI group, the overweight group, and the obesity group (all $P > 0.05$). 2) Cumulative pregnancy rates for AID cycles 1–6 were 17.60%, 31.60%, 43.08%, 54.37%, 61.83% and 73.68%, respectively. 3) Multivariate GEE analysis revealed that female age ($OR = 0.962$, 95% CI : 0.950–0.974, $P < 0.001$), endometrial thickness ($OR = 1.040$, 95% CI : 1.011–1.069, $P = 0.006$), and natural cycles ($OR = 1.171$, 95% CI : 1.060–1.294, $P = 0.002$) influenced clinical pregnancy rates. Compared with the normal BMI group, there were no statistically significant differences in clinical pregnancy rates of low BMI group, overweight group, and obese group (all $P > 0.05$). Multivariate logistic analysis showed that female age ($OR = 1.051$, 95% CI : 1.012–1.091, $P = 0.010$), endometrial thickness ($OR = 0.920$, 95% CI : 0.847–1.000, $P = 0.049$) and polycystic ovary syndrome ($OR = 1.927$, 95% CI : 1.044–3.556, $P = 0.036$) influenced spontaneous abortion rates. Compared with the normal BMI group, there were no statistically significant differences in spontaneous abortion rates of low BMI group, overweight group and obese group (all $P > 0.05$). 4) Cox regression analysis indicated that female age ($HR = 0.939$, 95% CI : 0.928–0.950, $P < 0.001$), endometrial thickness ($HR = 1.039$, 95% CI : 1.013–1.066, $P = 0.003$) and natural cycles ($HR = 1.957$, 95% CI : 1.785–2.146, $P < 0.001$) influenced cumulative pregnancy rates. Compared with the normal BMI group, there were no statistically significant differences in cumulative pregnancy rates of low BMI group, overweight group and obese group (all $P > 0.05$). **Conclusion** Female BMI does not significantly affect clinical pregnancy rates, spontaneous abortion rates and cumulative pregnancy rates in AID.

【Key words】 Artificial insemination with donor sperm; Clinical pregnancy rate; Spontaneous abortion rate; Cumulative pregnancy rate; Generalized estimating equations

临床研究

DOI: 10.3760/cma.j.cn101441-20250221-00084

收稿日期 2025-02-21 本文编辑 王李艳

引用本文: 黄园斐, 丁书芳, 林苏霞, 等. 拮抗剂方案新鲜胚胎移植后单用地屈孕酮片与联合用阴道黄体酮凝胶的临床结局比较 [J]. 中华生殖与避孕杂志, 2025, 45(8): 794-801. DOI: 10.3760/cma.j.cn101441-20250221-00084.

拮抗剂方案新鲜胚胎移植后单用地屈孕酮片与联合用阴道黄体酮凝胶的临床结局比较

黄园斐¹ 丁书芳¹ 林苏霞¹ 罗龙丹¹ 叶天民²

¹香港大学深圳医院生殖医学中心, 深圳 518053; ²上海交通大学医学院附属第六人民医院生殖医学中心, 上海 200233

通信作者: 叶天民, Email: drytm@163.com

【摘要】 目的 比较促性腺激素释放激素(gonadotropin-releasing hormone, GnRH)拮抗剂方案新鲜胚胎移植后口服地屈孕酮片与口服地屈孕酮片联合阴道黄体酮凝胶的临床结局。方法 采用回顾性队列研究, 分析2015年12月至2020年12月在香港大学深圳医院生殖医学中心进行的658个拮抗剂方案下的新鲜胚胎移植周期患者的临床资料, 每个周期均为患者首次行新鲜促排卵周期, 按照不同的黄体支持方案分为两组, 每天口服30 mg地屈孕酮片(记为A组, 368个周期), 每天口服20 mg地屈孕酮片并阴道置入90 mg黄体酮阴道缓释凝胶(记为B组, 290个周期)。对两组进行了1:1倾向性评分匹配以调整变量的混杂差异, 根据胚胎类型分层分为卵裂期胚胎移植周期及囊胚移植周期, 对两组的实验室结果及助孕结局进行对比。结果 经过倾向性评分匹配后, 两组之间的基线特征相似, 每组分别有251个周期进行了回顾性分析。匹配后, A组与B组的受精数[5(2, 7)比6(3, 9), $P=0.002$]、卵裂率[100.00%(86.31%, 100.00%)比87.28%(75.32%, 100.00%), $P<0.001$]、可用胚胎率[80.18%(54.64%, 100.00%)比67.48%(50.62%, 100.00%), $P=0.019$]的差异均有统计学意义; 其临床妊娠率、胚胎着床率、活产率、流产率、多胎妊娠率、卵巢过度刺激综合征(ovarian hyperstimulation syndrome, OHSS)

发生率和异位妊娠率组间比较差异均无统计学意义 (均 $P>0.05$)。根据胚胎类型分层研究, 卵裂期胚胎移植周期和囊胚移植周期中, A 组与 B 组在临床妊娠率、胚胎着床率、活产率、流产率、多胎妊娠率、OHSS 发生率和异位妊娠率方面差异均无统计学意义 (均 $P>0.05$)。结论 单独口服地屈孕酮片 30 mg 和口服地屈孕酮片 20 mg 联合阴道黄体酮凝胶在 GnRH 拮抗剂方案行新鲜胚胎移植周期中临床结局相当, 单用口服地屈孕酮片可以成为 GnRH 拮抗剂新鲜周期中黄体支持的新选择。

【关键词】 胚胎移植; 拮抗剂方案; 黄体支持; 地屈孕酮片; 阴道黄体酮凝胶

Comparison of clinical outcomes between oral dydrogesterone and vaginal progesterone gel plus oral dydrogesterone after fresh embryo transfer with GnRH antagonist protocol

Huang Yuanfei¹, Ding Shufang¹, Lin Suxia¹, Luo Longdan¹, Ye Tianmin²

¹ Center of Reproductive Medicine, the University of Hong Kong-Shenzhen Hospital, Shenzhen 518053, China; ² Center of Reproductive Medicine, Shanghai Sixth People's Hospital Affiliated to Shanghai Jiao Tong University School of Medicine, Shanghai 200233, China

Corresponding author: Ye Tianmin, Email: drytm@163.com

【Abstract】 Objective To compare the clinical outcomes of single oral dydrogesterone with vaginal progesterone gel plus oral dydrogesterone in gonadotropin-releasing hormone (GnRH) antagonist cycles with fresh embryo transfer. **Methods** This study retrospectively analyzed 658 treatment cycles of fresh embryo transfer cycle with GnRH antagonist protocol from December 2015 to December 2020 in the Center of Reproductive Medicine of the University of Hong Kong-Shenzhen Hospital. Each cycle was the first fresh stimulation cycle of the patients. The patients were divided into two groups according to different luteal support regimens. Group A included 368 cycles with a regimen of 30 mg dydrogesterone tablets orally daily, while group B included 290 cycles with a regimen of 90 mg vaginal progesterone gel vaginally daily combined with 20 mg dydrogesterone tablets orally daily. A 1 : 1 propensity score matching (PSM) was carried out to adjust for numerical differences and to balance between the two groups, and further they were divided into cleavage stage embryo transfer cycles and the blastocyst transfer cycles according to the different type of embryo for layer analysis, and the laboratory results and assisted reproductive outcomes of the two groups were compared. **Results** After matching, the baseline characteristics were comparable between the two groups, with 251 cycles remaining in each group for retrospective analysis. After PSM, statistically significant differences were observed between group A and group B in laboratory data including the number of fertilized oocytes [5 (2, 7) vs. 6 (3, 9), $P=0.002$], cleavage rate [100.0% (86.31%, 100.0%) vs. 87.28% (75.32%, 100.0%), $P<0.001$], and available embryo rate [80.18% (54.64%, 100.0%) vs. 67.48% (50.62%, 100.0%), $P=0.019$]. However, there were no significantly statistical differences in other laboratory data and clinical outcomes (all $P>0.05$). If we divided the data into two comparison according to the different type of embryo, there were no significantly statistical

differences in clinical pregnancy rate, embryo implantation rate, live birth rate, miscarriage rate, multiple pregnancy rate, ovarian hyperstimulation syndrome incidence, and ectopic pregnancy rate neither in day 2 cleavage stage embryo transfer cycles nor in the blastocyst transfer cycles. **Conclusion** In this study, the clinical outcomes are similar between taking 30 mg of dydrogesterone tablets orally alone and taking 20 mg of dydrogesterone tablets orally combined with vaginal progesterone gel in the fresh embryo transfer cycle of the GnRH antagonist protocol. Moreover, taking dydrogesterone tablets orally alone can be a new option for luteal support in the fresh cycle of the GnRH antagonist protocol.

【Key words】 Embryo transfer; Antagonist protocol; Luteal phase support; Dydrogesterone; Vaginal progesterone gel

临床研究

DOI: 10.3760/cma.j.cn101441-20250319-00130

收稿日期 2025-03-19 本文编辑 孙敏

引用本文: 郭小妮, 刘晓洁, 张孝东, 等. 优质胚胎冷冻保存时长对临床妊娠结局影响的回顾性队列研究[J]. 中华生殖与避孕杂志, 2025, 45(8): 802-808. DOI: 10.3760/cma.j.cn101441-20250319-00130.

($OR=1.053$, 95% CI : $0.945\sim1.174$, $P=0.350$)、活产率($OR=1.061$, 95% CI : $0.958\sim1.175$, $P=0.255$)均无影响, 7~12个月组(临床妊娠率 $OR=0.706$, 95% CI : $0.619\sim0.806$, $P<0.001$; 活产率 $OR=0.748$, 95% CI : $0.658\sim0.850$, $P<0.001$)和>12个月组(临床妊娠率 $OR=0.611$, 95% CI : $0.535\sim0.699$, $P<0.001$; 活产率 $OR=0.627$, 95% CI : $0.550\sim0.714$, $P<0.001$)

胚胎数、胚胎发育阶段、获卵数、行胚胎植入前遗传学检测比例差异均有统计学意义(均 $P<0.05$)。≤3个月组、4~6个月组、7~12个月组和>12个月组的临床妊娠率分别为69.04% (2 092/3 030)、70.15% (2 240/3 193)、61.16% (896/1 465)和57.69% (750/1 300), 活产率分别为58.58% (1 775/3 030)、60.04% (1 917/3 193)、51.40% (753/1 465)和47.00% (611/1 300)。

调整混杂因素后, 与≤3个月组相比, 4~6个月组对临床妊娠率($OR=0.982$, 95% CI : $0.874\sim1.103$, $P=0.754$)、活产率($OR=0.989$, 95% CI : $0.887\sim1.102$, $P=0.835$)均无影响, 7~12个月组(临床妊娠率 $OR=0.772$, 95% CI : $0.671\sim0.888$, $P<0.001$; 活产率 $OR=0.805$, 95% CI : $0.704\sim0.921$, $P=0.002$)和>12个月组(临床妊娠率 $OR=0.765$, 95% CI : $0.662\sim0.885$, $P<0.001$; 活产率 $OR=0.772$, 95% CI : $0.671\sim0.888$, $P<0.001$)的临床妊娠率和活产率显著下降。早产率及流产率在4组间差异均无统计学意义(均 $P>0.05$)。

优质胚胎冷冻保存时长对临床妊娠结局影响的回顾性队列研究

郭小妮 刘晓洁 张孝东 黄国宁 张琦

重庆市妇幼保健院 重庆医科大学附属妇女儿童医院生殖医学中心

国家卫生健康委出生缺陷与生殖健康重点实验室, 重庆 400013

通信作者: 张琦, Email: zgqi0613@163.com

【摘要】 目的 评估优质胚胎冷冻保存时长对临床妊娠结局的影响。**方法** 采用回顾性队列研究, 分析 2016 年 1 月至 2023 年 12 月在重庆市妇幼保健院生殖医学中心接受人工周期内膜准备并进行优质胚胎移植的 8 988 个冻融胚胎移植周期。根据胚胎冷冻保存时长分为 4 组: ≤ 3 个月组 ($n=3\ 030$)、4~6 个月组 ($n=3\ 193$)、7~12 个月组 ($n=1\ 465$)、 >12 个月组 ($n=1\ 300$)。根据 Istanbul 共识和 Gardner 评分选择优质卵裂期胚胎和囊胚期胚胎。优质卵裂期胚胎定义为评级 ≤ 2 级的胚胎, 优质囊胚定义为评级 ≥ 4 BB。采用广义估计方程进行多因素分析, 主要结局指标为临床妊娠率, 次要结局指标为活产率、流产率及早产率。结果 4 组间年龄、体质指数、抗苗勒管激素、授精方式、胚胎移植日子宫内膜厚度、不孕因素、不孕类型、移植胚胎数、胚胎发育阶段、获卵数、行胚胎植入前遗传学检测比例差异均有统计学意义 (均 $P<0.05$)。 ≤ 3 个月组、4~6 个月组、7~12 个月组和 >12 个月组的临床妊娠率分别为 69.04% (2 092/3 030)、70.15% (2 240/3 193)、61.16% (896/1 465) 和 57.69% (750/1 300), 活产率分别为 58.58% (1 775/3 030)、60.04% (1 917/3 193)、51.40% (753/1 465) 和 47.00% (611/1 300), 4 组间比较差异均有统计学意义 (均 $P<0.001$)。调整混杂因素后, 与 ≤ 3 个月组相比, 4~6 个月组对临床妊娠率 ($OR=0.982$, 95% CI : 0.874~1.103, $P=0.754$)、活产率 ($OR=0.989$, 95% CI : 0.887~1.102, $P=0.835$) 均无影响, 7~12 个月组 (临床妊娠率 $OR=0.772$, 95% CI : 0.671~0.888, $P<0.001$; 活产率 $OR=0.805$, 95% CI : 0.704~0.921, $P=0.002$) 和 >12 个月组 (临床妊娠率 $OR=0.765$, 95% CI : 0.662~0.885, $P<0.001$; 活产率 $OR=0.772$, 95% CI : 0.671~0.888, $P<0.001$) 的临床妊娠率和活产率显著下降。早产率及流产率在 4 组间差异均无统计学意义 (均 $P>0.05$)。根据年龄分层, 结果与总人群一致。结论 优质胚胎玻璃化冷冻时长超过 6 个月与临床妊娠率及活产率呈负相关, 胚胎冷冻保存时长应作为选择移植胚胎的考量因素之一。

【关键词】 玻璃化; 冻融胚胎移植; 冷冻时长; 临床妊娠率; 活产率

基金项目: 国家自然科学基金 (82371728); 重庆市科技局创新与应用发展专项重点项目 (CSTB2022TIAD-KPX0146); 重庆市卫健委妇幼临床项目 (2023FY102)

Impact of cryostorage duration on clinical outcomes: a retrospective cohort study of vitrified high-quality embryos

Guo Xiaoni, Liu Xiaojie, Zhang Xiaodong, Huang Guoning, Zhang Qi

Center for Reproductive Medicine, Chongqing Maternal and Child Healthcare Hospital; Affiliated Women's and Children's Hospital of Chongqing Medical University; NHC Key Laboratory of Birth Defects and Reproductive Health, Chongqing 400013, China

Corresponding author: Zhang Qi, Email: zgqi0613@163.com

【 Abstract 】 **Objective** To evaluate the effect of prolonged cryopreservation duration of high-quality embryos on clinical outcomes. **Methods** A retrospective cohort study was conducted, analyzing 8 988 frozen-thawed embryo transfer cycles performed from January 2016 to December 2023 at the Center for Reproductive Medicine, Chongqing Maternal and Child Healthcare Hospital where patients underwent endometrial preparation with artificial cycles and subsequent transfer of high-quality embryos. Embryos were divided into four groups according to the length of time they had been cryopreserved: ≤ 3 -month group ($n=3\ 030$), 4–6-month group ($n=3\ 193$), 7–12-month group ($n=1\ 465$), and >12 -month group ($n=1\ 300$). High-quality cleavage-stage embryos and blastocysts were selected according to the Istanbul Consensus and Gardner grading system. High-quality cleavage-stage embryos were defined as those graded ≤ 2 , while high-quality blastocysts were defined as those graded ≥ 4 BB. Generalized estimating equations were employed for multivariate analysis. Primary outcome indicator was clinical pregnancy rate, with secondary outcome indicators comprising live birth rate, miscarriage rate and preterm birth rate. **Results** Significant intergroup differences were observed in baseline characteristics, including age, body mass index, anti-Müllerian hormone levels, fertilization method, endometrial thickness on transfer day, infertility etiology, infertility type, number of embryos transferred, embryo culture duration, number of eggs obtained, and preimplantation genetic testing (all $P<0.05$). Clinical pregnancy rates for the ≤ 3 -month, 4–6-month, 7–12-month, and >12 -month groups were 69.04% (2 092/3 030), 70.15% (2 240/3 193), 61.16% (896/1 465), and 57.69% (750/1 300), respectively, and live birth rates were 58.58% (1 775/3 030), 60.04% (1 917/3 193), 51.40% (753/1 465), and 47.00% (611/1 300), with significantly differences (all $P<0.001$). After adjusting for confounders via multivariate analysis, the 4–6-month group showed no statistically significant difference in clinical pregnancy rate or live birth rate compared with the ≤ 3 -month group (clinical pregnancy: $OR=0.982$, 95% CI : 0.874–1.103, $P=0.754$; live birth: $OR=0.989$, 95% CI : 0.887–1.102, $P=0.835$). However, both the 7–12-month group (clinical pregnancy: $OR=0.772$, 95% CI : 0.671–0.888, $P<0.001$; live birth: $OR=0.805$, 95% CI : 0.704–0.921, $P=0.002$) and >12 -month group (clinical pregnancy: $OR=0.765$, 95% CI : 0.662–0.885, $P<0.001$; live birth: $OR=0.772$, 95% CI : 0.671–0.888, $P<0.001$) exhibited significant decreases in clinical pregnancy rate and live birth rate. No significant differences were observed in miscarriage rate and preterm birth rate among the four groups (all $P>0.05$). Stratified by age, the results were consistent with the total population. **Conclusion** The duration of high-quality embryo vitrification freezing exceeding 6 months is negatively correlated with clinical pregnancy rate and live birth rate, and cryostorage time should be considered as a relevant factor in embryo selection.

【 Key words 】 Vitrification; Frozen-thawed embryo transfer; Cryopreservation duration; Clinical pregnancy rate; Live birth rate

Fund program: National Natural Science Foundation of China (82371728); Chongqing Science and Technology Commission Technological Innovation and

实验研究

DOI: 10.3760/cma.j.cn101441-20250219-00077

收稿日期 2025-02-19 本文编辑 李天琦

引用本文: 刘超, 程兰兰, 谭丽. 脐带血单个核细胞宫腔灌注对薄型子宫内膜建模小鼠子宫内膜厚度及容受性的影响 [J]. 中华生殖与避孕杂志, 2025, 45(8): 809-817. DOI: 10.3760/cma.j.cn101441-20250219-00077.

脐带血单个核细胞宫腔灌注对薄型子宫内膜建模小鼠子宫内膜厚度及容受性的影响

刘超¹ 程兰兰¹ 谭丽²

¹郑州大学第二附属医院生殖医学部, 郑州 450014; ²成都双楠医院

生殖医学中心, 成都 610041

通信作者: 谭丽, Email: litan668@126.com

【摘要】 目的 探究脐带血单个核细胞 (umbilical cord blood mononuclear cells, UCB-MNCs) 宫腔灌注对薄型子宫内膜的治疗作用及相关机制。方法 选用 SPF 级 6~8 周龄昆明小鼠, 采用 95%乙醇宫腔灌注 5 min 建立薄型子宫内膜模型。采用完全随机化分组法, 将 40 只动情周期规律的雌性小鼠随机分为 4 组: 未处理组 (无干预, $n=10$)、假手术组 (动情期模拟手术: 开腹, 分离出子宫后关腹, $n=10$)、实验组 (造模后间隔 1 个动情周期, 于动情期双侧宫腔灌注 UCB-MNCs, $n=10$) 和阴性对照组 (造模后间隔 1 个动情周期, 于动情期双侧宫腔灌注生理盐水, $n=10$)。灌注 UCB-MNCs/生理盐水后间隔 2 个动情周期, 于动情期收集所有组别子宫组织。通过 HE 染色观察子宫内膜形态学变化, 测量内膜厚度并计数腺体数量。采用 Western blotting、逆转录实时荧光定量聚合酶链式反应检测各组子宫内膜白血病抑制因子 (leukemia inhibitory factor, LIF)、血管内皮生长因子 (vascular endothelial growth factor, VEGF)、整合素 (integrin, ITG) α_v 、ITG β_3 、增殖细胞核抗原 (proliferating cell nuclear antigen, PCNA) 蛋白及 mRNA 的相对表达量, 并进行组间比较。结果 未处理组子宫内膜厚度 [(507.32 $\pm$ 85.66)

μm]、腺体数量 [(18.67±6.66) 个] 与假手术组 [(502.78±73.26) μm 、(19.33±7.73) 个] 比较, 差异均无统计学意义 (均 $P>0.05$)。实验组子宫内膜厚度 [(347.71±82.24) μm] 及腺体数量 [(15.00±2.65) 个] 显著高于阴性对照组 [(118.85±29.19) μm , $P<0.001$; (2.00±2.00) 个, $P=0.030$]。未处理组与假手术组间子宫内膜 LIF、VEGF、ITG α_v 、ITG β_3 、PCNA 的蛋白及 mRNA 相对表达量差异均无统计学意义 (均 $P>0.05$)。实验组子宫内膜 LIF、VEGF、ITG α_v 、ITG β_3 、PCNA 的蛋白及 mRNA 相对表达量均高于阴性对照组 (均 $P<0.05$)。结论 UCB-MNCs 宫腔灌注可能通过上调 PCNA、LIF、VEGF、ITG α_v 、ITG β_3 的表达, 促进子宫内膜的再生修复, 并改善子宫内膜容受性。

【关键词】 脐带血单个核细胞; 薄型子宫内膜; 子宫内膜厚度; 子宫内膜容受性

Impact of umbilical cord blood mononuclear cells intrauterine perfusion on endometrial thickness and endometrial receptivity in mice with thin endometrium

Liu Chao¹, Cheng Lanlan¹, Tan Li²

¹ Department of Reproductive Medicine, Second Affiliated Hospital of Zhengzhou University, Zhengzhou 450014, China; ² Reproductive Medicine Center, Chengdu Shuangnan Hospital, Chengdu 610041, China

Corresponding author: Tan Li, Email: litan668@126.com

【Abstract】 **Objective** To investigate the therapeutic effects and underlying mechanisms of intrauterine perfusion of umbilical cord blood mononuclear cells (UCB-MNCs) on thin endometrium. **Methods** SPF-grade Kunming mice aged 6–8 weeks were selected. A mouse model of thin endometrium was established by infusing 95% ethanol into the uterine cavity for a duration of 5 min. Using a completely randomized grouping method, 40 female mice with regular estrous cycles were randomly divided into four groups: untreated group (no intervention, $n=10$), sham-operated group (operation without modeling, $n=10$), experimental group (intrauterine infusion of UCB-MNCs during estrus after one estrous cycle post-modeling, $n=10$) and negative control group (intrauterine infusion of saline during estrus after one estrous cycle post-modeling, $n=10$). Following the administration of UCB-MNCs or physiological saline, all groups' uterine tissues were collected two estrous cycles later during their respective estrus phases. Hematoxylin-eosin staining was used to assess endometrial morphology, measure thickness, and count glands. Western blotting and reverse transcription real-time quantitative polymerase chain reaction were utilized to measure the relative protein and mRNA expression levels of leukemia inhibitory factor (LIF), vascular endothelial growth factor (VEGF), integrin (ITG) α_v , ITG β_3 and proliferating cell nuclear antigen (PCNA) in the endometrium across different groups for intergroup comparisons. **Results** The endometrial thickness and the number of glands in the untreated group [(507.32±85.66) μm , 18.67±6.66] showed no statistically significant differences compared with those in the sham-operated group [(502.78±73.26) μm , 19.33±7.73, all $P>0.05$]. The experimental group showed significantly increased endometrial thickness [(347.71±82.24) μm vs.

(118.85±29.19) μm , $P<0.001$] and gland number (15.00±2.65 vs. 2.00±2.00, $P=0.030$) compared with the negative control group. There was no statistically significant difference in the relative protein and mRNA expression levels of LIF, VEGF, ITG α_v , ITG β_3 , and PCNA in the endometrium between the untreated group and the sham-operated group (all $P>0.05$). The relative protein and mRNA expression levels of endometrial LIF, VEGF, ITG α_v , ITG β_3 and PCNA of the experimental group were all significantly higher than those in the negative control group (all $P<0.05$). **Conclusion** Intrauterine perfusion with UCB-MNCs may promote endometrial regeneration and repair, as well as improve endometrial receptivity, through the upregulation of the expression levels of PCNA, LIF, VEGF, and ITG α_v , ITG β_3 .

【Key words】 Umbilical cord blood mononuclear cells; Thin endometrium; Endometrial thickness; Endometrial receptivity

循证医学

DOI: 10.3760/cma.j.cn101441-20250305-00105

收稿日期 2025-03-05 本文编辑 王李艳

引用本文: 高媛, 宋再伟, 姜丹, 等. 体外受精促排卵领域指南与共识的现状分析与质量评价[J]. 中华生殖与避孕杂志, 2025, 45(8): 818-825. DOI: 10.3760/cma.j.cn101441-20250305-00105.

体外受精促排卵领域指南与共识的现状分析与质量评价

高媛¹ 宋再伟¹ 姜丹¹ 杨蕊² 黄育文¹ 李蓉² 赵荣生¹

¹北京大学第三医院药学部 北京大学医学部药物评价中心, 北京

100191; ²北京大学第三医院妇产科生殖医学中心, 北京 100191

通信作者: 赵荣生, Email: zhaorongsheng@bjmu.edu.cn

【摘要】 目的 系统评价体外受精促排卵领域指南与共识的制订现状及方法学质量, 为后续相关指南制定与研究开展提供参考。方法 系统检索PubMed、Embase、CNKI、Wangfang、Sinomed 等数据库及指南相关网站, 检索时间为建库至 2024 年 10 月 31 日。对符合纳入标准的文献进行资料提取, 并采用指南研究和评估工具 II (Appraisal of Guidelines for Research and Evaluation II, AGREE II) 进行方法学质量评估。采用描述性分析方法, 并以可视化数据图呈现结果。结果 共纳入 23 项研究, 其中 8 项为指南, 15 项为专家共识。21 项 (91.3%) 研究关注促

排卵方案选择,多数聚焦在首次行 IVF 治疗患者的促排卵方案;12 项 (52.2%) 研究关注促排卵中促性腺激素起始剂量的选择。方法学方面,在 AGREE II 的 6 个评估领域中,各领域的标准化平均得分率分别为范围和目的 (85.1%)、参与人员 (66.6%)、严谨性 (36.5%)、清晰性 (70.1%)、应用性 (36.4%) 及独立性 (46.2%)。结论 当前体外受精促排卵领域的推荐意见存在差异与空白、方法学质量不齐,一定程度上阻碍了相关证据向临床实践的转化。未来应围绕促排卵方案与促性腺激素起始剂量的选择进一步开展研究,推动该领域指南共识的更新完善以指导促排卵精准治疗。

【关键词】 体外受精; 促排卵; 临床指南; 专家共识; 系统综述

基金项目: 国家自然科学基金 (72304007、72474013); 北京市卫生健康科技成果和适宜技术推广项目 (BHTPP2024007); 北京大学第三医院—丽珠医药创新药物联合研发中心资助项目

Current situation analysis and quality evaluation of guidelines and consensus in ovarian stimulation for *in vitro* fertilization

Gao Yuan¹, Song Zaiwei¹, Jiang Dan¹, Yang Rui², Huang Yuwen¹, Li Rong², Zhao Rongsheng¹

¹ Department of Pharmacy, Peking University Third Hospital; Institute for Drug Evaluation of Peking University Health Science Center, Beijing 100191, China; ² Center for Reproductive Medicine, Department of Obstetrics and Gynecology, Peking University Third Hospital, Beijing 100191, China

Corresponding author: Zhao Rongsheng, Email: zhaorongsheng@bjmu.edu.cn

【Abstract】 Objective To systematically evaluate the current status and methodological quality of guideline and consensus in ovarian stimulation for *in vitro* fertilization (IVF), and to provide reference for the development of future guidelines and research. **Methods** A systematic search was conducted in databases including PubMed, Embase, CNKI, Wanfang, Sinomed and relevant guideline websites from inception to October 2024. Data were extracted from the literature that met the inclusion criteria, and methodological quality was assessed using the Appraisal of Guidelines for Research and Evaluation II (AGREE II) tool. Descriptive analysis was performed with results presented in visual data charts. **Results** A total of 23 studies were included, with 8 guidelines and 15 consensus. Totally 21 (91.3%) studies focused on the selection of ovarian stimulation protocols, with the majority concentrating on protocols for patients undergoing IVF treatment for the first cycle, and 12 (52.2%) studies focused on the selection of gonadotropin starting dosage. In terms of methodology, the average standardized scores in the 6 assessment domains of the AGREE II tool were as follows: scope and purpose (85.1%), stakeholder involvement (66.6%), rigour of development (36.5%), clarity of presentation (70.1%), applicability (36.4%), and editorial independence (46.2%). **Conclusion** Current recommendations in ovarian stimulation for IVF exhibit inconsistencies, and the methodological quality varies, which partially hinders the translation of evidence into clinical practice. Future research should further focus on the selection of ovarian stimulation protocols and

gonadotropin starting dosage, promoting the update and refinement of guidelines and consensus to guide the precise treatment of ovarian stimulation.

【Key words】 *In vitro* fertilization; Ovarian stimulation; Clinical guidelines; Expert consensus; Systematic review

Fund program: National Natural Science Foundation of China (72304007, 72474013); Beijing Municipal Health Science and Technology Achievements and Appropriate Technology Promotion Project (BHTPP2024007); Joint Research and Development Project of Innovative Drugs by Peking University Third Hospital and Livzon Pharmaceutical Group

循证医学

DOI: 10.3760/cma.j.cn101441-20250311-00117

收稿日期 2025-03-11 本文编辑 李天琦

引用本文: 侯志金, 刘洋, 杨晓玲, 等. *CYP17A1* 基因突变所致先天性肾上腺皮质增生症合并不孕症患者的最佳助孕治疗策略: 一项系统综述 [J]. 中华生殖与避孕杂志, 2025, 45(8): 826-832. DOI: 10.3760/cma.j.cn101441-20250311-00117.

CYP17A1 基因突变所致先天性肾上腺皮质增生症合并不孕症患者的最佳助孕治疗策略: 一项系统综述

侯志金 刘洋 杨晓玲 李东娅 孟昱时

昆明医科大学第二附属医院生殖医学科, 昆明 650000

通信作者: 孟昱时, Email: mengyushi0102@163.com

【摘要】 **目的** 综合评估 *CYP17A1* 缺陷所致先天性肾上腺皮质增生症 [包括 17 α -羟化酶/17, 20-裂解酶缺乏症 (17 α -hydroxylase/17, 20-lyase deficiency, 17-OHD) 及孤立性 17, 20-裂解酶缺乏症 (isolated 17, 20-lyase deficiency, IDL)] 合并不孕症患者接受助孕治疗的结局及策略。**方法** 通过系统检索 PubMed、Embase、Web of Science、Ovid、中国知网、万方数据库和中国生物医学文献数据库, 提取数据包括患者基本信息、治疗方案及妊娠结局, 并进行描述性分析。结果纳入 19 篇文献, 涵盖 23 例成功活产的病例。23 例成功活产患者中, 17-OHD 患者 19 例, 其中 17 例通过体外受精-胚胎移植 (*in vitro* fertilization and embryo transfer, IVF-ET) 妊娠, 1 例自然妊娠, 1 例赠卵妊娠; IDL 患者 4 例, 其中 2 例自然妊娠, 2 例通过 IVF-ET 妊娠。**结论** IVF-ET 是部分型 17-OHD 合并不孕患者

助孕的首选方案，而有自然排卵的 IDL 合并不孕患者可尝试自然妊娠。糖皮质激素在改善妊娠结局中具有关键作用，但其使用需个体化。

【关键词】 *CYP17A1* 基因突变； 17 α -羟化酶/17, 20-裂解酶缺乏症； 孤立性 17, 20-裂解酶缺乏症； 助孕

基金项目：云南省“万人计划”名医专项人才项目（云卫人发〔2019〕35号）

Optimal assisted reproductive strategies for infertile patients with congenital adrenal hyperplasia associated with *CYP17A1* gene mutations: a systematic review

Hou Zhijin, Liu Yang, Yang Xiaoling, Li Dongya, Meng Yushi

Department of Reproductive Medicine, the Second Affiliated Hospital of Kunming Medical University, Kunming 650000, China

Corresponding author: Meng Yushi, Email: mengyushi0102@163.com

【Abstract】 Objective To comprehensively evaluate treatment outcomes and assisted reproductive approaches in infertility patients with *CYP17A1*-deficient congenital adrenal hyperplasia [including 17 α -hydroxylase/17,20-lyase deficiency (17-OHD) and isolated 17,20-lyase deficiency (IDL)]. **Methods** In this study, PubMed, Embase, Web of Science, Ovid, CNKI, Wanfang Data base, and the Chinese Biomedical Literature Database were systematically searched. The data extracted included basic patient information, treatment plans, and pregnancy outcomes, which were then analyzed descriptively. **Results** Totally 19 articles were included, covering 23 cases of successful live births. Among the 23 patients with successful live births, there were 19 patients with 17-OHD, including 17 cases became pregnant through *in vitro* fertilization and embryo transfer (IVF-ET), 1 case through natural pregnancy, and 1 case through oocyte donation pregnancy; there were 4 cases with IDL, including 2 cases through natural pregnancy and 2 cases through IVF-ET pregnancy. **Conclusion** IVF-ET is the preferred fertility treatment option for patients with partial form of 17-OHD. For IDL patients who have natural ovulation, natural pregnancy can be attempted. Glucocorticoids play a crucial role in improving pregnancy outcomes, but their use needs to be individualized.

【 Key words 】 *CYP17A1* gene mutation; 17 α -hydroxylase/17,20-lyase deficiency; Isolated 17, 20-lyase deficiency; Fertility treatment

Fund program: Famous Doctor Special Talent Project of the "Ten Thousand Talents Plan" in Yunnan Province (Yunwei Renfa [2019] No. 35)

·个案报道·

DOI: 10.3760/cma.j.cn101441-20250227-00093

收稿日期 2025-02-27 本文编辑 宋培培

引用本文：邓杰，马宁，周多，等. 基于二代测序的 PGT 技术偶然发现隐匿性复杂染色体重排男性携带者 1 例报道 [J]. 中华生殖与避孕杂志，2025，45(8): 833-836. DOI: 10.3760/cma.j.cn101441-20250227-00093.

基于二代测序的 PGT 技术偶然发现隐匿性复杂染色体重排男性携带者 1 例报道

邓杰¹ 马宁² 周多¹ 王火鸟¹ 周俏苗¹

¹海南省妇女儿童医学中心医学遗传与产前诊断科，海口 570206；²

海南省妇女儿童医学中心生殖医学中心，海口 570206

通信作者：周俏苗，Email: zhouqiaomiao@126.com

【摘要】 本文报道 1 例基于二代测序 (next-generation sequencing, NGS) 的植入前遗传学检测 (preimplantation genetic testing, PGT) 技术偶然发现隐匿性复杂染色体重排 (complex chromosomal rearrangement, CCR) 男性携带者的病例并进行文献复习。该病例因核型为 46, XY, t (8; 14) (p21; q13) 而选择 PGT 助孕，胚胎的异常拷贝数变异 (copy number variation, CNV) 结果提示男方 8 号染色体可能存在 2 个不同的断裂点，推测其可能存在除了已知易位以外的其他结构异常。经改良高分辨 G 显带技术对夫妇外周血淋巴细胞分析证实了这一发现，最终确定该病例核型为 46, XY, der (8) inv (8) (p22q12.2) t (8; 14) (p22; q21.3), der (14) t (8; 14)。改良高分辨 G 显带技术可提高隐匿性 CCRs 的检出率，减少漏诊。本案例表明，通过对胚胎异常 CNV 结果的 PGT 分析，可能发现此前未检出的染色体异常，从而为患者提供更精准的遗传咨询和生殖决策支持。

【关键词】 植入前遗传学检测；二代测序；G 显带；复杂染色体重排；高分辨

基金项目：海南省重大科技计划项目 (ZDKJ2021037)；海南省优秀人才团队 (琼人才办通 [2021] 21 号)；海南省临床医学中心建设项目 (琼卫医函 [2021] 75 号)

Incidental identification of a cryptic complex chromosomal rearrangement in a male carrier by next-generation sequencing-based PGT: a case report

Deng Jie¹, Ma Ning², Zhou Duo¹, Wang Huoniao¹, Zhou Qiaomiao¹

¹ Department of Genetics and Prenatal Diagnosis, Hainan Women and Children's Medical Center, Haikou 570206, China; ² Reproductive Medical Center, Hainan Women and Children's Medical Center, Haikou 570206, China

Corresponding author: Zhou Qiaomiao, Email: zhouqiaomiao@126.com

【Abstract】 This study presents a case of a male carrier with a cryptic complex chromosomal rearrangement (CCR) incidentally identified by

next-generation sequencing (NGS)-based preimplantation genetic testing (PGT), along with a literature review. The couple underwent PGT because of the male partner's known balanced translocation karyotype [46,XY,t(8;14)(p21;q13)]. Embryonic copy number variation (CNV) analysis unexpectedly revealed two distinct breakpoints on chromosome 8 in the male partner, suggesting additional structural abnormalities beyond the documented translocation. Modified high-resolution G-banding analysis of peripheral blood lymphocytes confirmed these findings. The male partner's high-resolution karyotype was ultimately characterized as 46,XY,der(8)inv(8)(p22q12.2)t(8;14)(p22;q21.3),der(14)t(8;14). The modified high-resolution G-banding technique demonstrated enhanced detection of cryptic CCRs, reducing diagnostic oversight. This case demonstrates that PGT analysis of abnormal embryonic CNV results can uncover previously undetected chromosomal abnormalities, thereby improving genetic counseling and reproductive decision-making.

【Key words】 Preimplantation genetic testing; Next-generation sequencing; G banding; Complex chromosome rearrangements; High resolution

Fund programs: Major Science and Technology Program of Hainan Province (ZDKJ2021037); Excellent Talent Team of Hainan Province (QRCBT [2021] No. 21); Project Supported by Hainan Province Clinical Medical Center (QWYH [2021] No. 75)

综述

DOI: 10.3760/cma.j.cn101441-20250317-00124

收稿日期 2025-03-17 本文编辑 王李艳

引用本文: 余璐萍, 梅洁. 人工智能在子宫腺肌病精准化诊疗中的研究进展[J]. 中华生殖与避孕杂志, 2025, 45(8): 837-840. DOI: 10.3760/cma.j.cn101441-20250317-00124.

人工智能在子宫腺肌病精准化诊疗中的研究进展

余璐萍 梅洁

南京大学医学院附属鼓楼医院生殖医学中心, 南京 210008

通信作者: 梅洁, Email: meijie560@163.com

【摘要】 子宫腺肌病是一种常见的育龄期女性妇科疾病, 以月经过多、痛经及不孕为主要临床特征, 严重影响患者生活质量, 并增加医疗负担。由于其临床表现缺乏特异性, 传统影像学诊断依赖于操作者经验, 且不同患者的病变范围和严重

程度存在较大异质性，导致疾病的早期识别和精准分型面临挑战。近年来，人工智能 (artificial intelligence, AI) 在医学影像分析、病理学评估及个性化治疗方案制定等方面展现出重要应用价值。基于深度学习的计算机辅助诊断技术能够自动识别超声及磁共振成像中的子宫腺肌病特征，提高诊断的准确性和一致性。此外，AI 在预测治疗反应及优化个体化管理策略方面亦取得了初步进展。本文对 AI 在子宫腺肌病诊疗中应用的研究进展进行综述，重点探讨其在优化诊断流程、提升临床决策效率及推进个体化治疗中的应用价值。

【关键词】 子宫腺肌病； 人工智能； 计算机辅助诊断； 深度学习； 个性化诊疗

基金项目：国家自然科学基金 (82301891)

Research progress of artificial intelligence in the precision diagnosis and treatment of adenomyosis

Yu Luping, Mei Jie

Reproductive Medicine Center, Nanjing Drum Tower Hospital, the Affiliated Hospital of Nanjing University Medical School, Nanjing 210008, China

Corresponding author: Mei Jie, Email: meijie560@163.com

【Abstract】 Adenomyosis is a common gynecological disease in women of reproductive age, characterized by menorrhagia, dysmenorrhea, and infertility. It significantly impairs patients' quality of life and imposes a substantial healthcare burden. Due to its nonspecific clinical manifestations, traditional imaging diagnosis relies heavily on operator experience, and the heterogeneity in lesion distribution and severity among patients poses challenges to early detection and precise classification. Recent advances in artificial intelligence (AI) have demonstrated significant potential in medical imaging analysis, pathological assessment, and personalized treatment planning. Deep learning-based computer-aided diagnostic techniques can automatically identify adenomyosis features in ultrasound and magnetic resonance imaging, thereby improving diagnostic accuracy and consistency. Moreover, AI has shown emerging utility in predicting treatment responses and tailoring individualized management strategies. This review summarizes current AI applications in the diagnosis and treatment of adenomyosis, emphasizing its value in streamlining diagnostic workflows, enhancing clinical decision-making, and advancing precision medicine.

【 Key words 】 Adenomyosis; Artificial intelligence; Computer-aided diagnosis; Deep learning; Personalized treatment

Fund program: National Natural Science Foundation of China (82301891)

综述

DOI: 10.3760/cma.j.cn101441-20250220-00080

收稿日期 2025-02-20 本文编辑 孙敏

引用本文：曹芸蕾，杜晶. 中性粒细胞在子宫内膜异位症中的研究进展[J]. 中华生殖与避孕杂志, 2025, 45(8): 841-845. DOI: 10.3760/cma.j.cn101441-20250220-00080.

中性粒细胞在子宫内膜异位症中的研究进展

曹芸蕾¹ 杜晶²

¹复旦大学附属妇产科医院普妇科, 上海 200011; ²上海市生物医药

技术研究院 上海市疾病与健康基因组学重点实验室 国家卫生健

康委生育调节药械重点实验室, 上海 200237

通信作者: 杜晶, Email: jing.du@sibpt.cn

【摘要】 子宫内膜异位症 (endometriosis, EMS) 是临床上常见的妇科疾病之一, 主要导致痛经、慢性盆腔痛和不孕。EMS 在育龄女性中的发病率高达 10%, 给女性的身心健康带来了极大危害。有关 EMS 的发病机制众说纷纭, 迄今尚无一种假说能很好地解释其发生。大量研究证明 EMS 患者的腹腔内存在慢性炎症反应, 多种免疫细胞和细胞因子参与了这一过程。因此, 免疫因素在 EMS 的发生、发展中所起的作用成为了最近研究的热点。而中性粒细胞作为炎症反应过程中主要参与者, 在 EMS 的研究当中受到了广泛关注。本文将就中性粒细胞在 EMS 中的研究进展作一综述。

【关键词】 子宫内膜异位症; 免疫学; 细胞因子; 中性粒细胞; 中性粒细胞胞外陷阱

Research progress of neutrophils in endometriosis

Cao Yunlei¹, Du Jing²

¹ Department of General Gynecology, Shanghai Obstetrics and Gynecology Hospital, Fudan University, Shanghai 200011, China; ² Shanghai-MOST Key Laboratory of Health and Disease Genomics; NHC Key Lab of Reproduction Regulation; Shanghai Institute for Biomedical and Pharmaceutical Technologies, Shanghai 200237, China
Corresponding author: Du Jing, Email: jing.du@sibpt.cn

【Abstract】 Endometriosis (EMS) is one of the most common clinical gynecological diseases, which mainly causes dysmenorrhea, chronic pelvic pain and infertility. The incidence of EMS in women of childbearing age is as high as 10%, which brings great harm to women's physical and mental health. There are many theories about the pathogenesis of EMS. So far, no single hypothesis can fully explain its occurrence. Numerous studies have demonstrated chronic inflammatory reactions in the peritoneal cavity of patients with EMS, and multiple immune cells and cytokines are involved in this process. Therefore, the role of immune factors in the occurrence and development of EMS has become the focus of recent research.

Neutrophils, as the main participants in the process of inflammatory response, have received wide attention in the study of endometriosis. This paper reviews the research progress on neutrophils in EMS.

【 Key words 】 Endometriosis; Immunology; Cytokine; Neutrophils; Neutrophil extracellular traps

综述

DOI: 10.3760/cma.j.cn101441-20250206-00049

收稿日期 2025-02-06 本文编辑 孙敏

引用本文: 黄澳淼, 张印峰, 刘聪, 等. 无创胚胎植入前非整倍体检测技术的研究进展及展望[J]. 中华生殖与避孕杂志, 2025, 45(8): 846-850. DOI: 10.3760/cma.j.cn101441-20250206-00049.

无创胚胎植入前非整倍体检测技术的研究进展及展望

黄澳淼¹ 张印峰² 刘聪² 韩莹² 罗海宁²

¹南开大学医学院, 天津 300071; ²天津市中心妇产科医院 南开大学附属妇产科医院生殖医学中心, 天津 300100

通信作者: 罗海宁, Email: 30317012@nankai.edu.cn

【摘要】 胚胎植入前非整倍体检测 (preimplantation genetic testing for aneuploidies, PGT-A) 技术是目前高龄 (年龄 ≥ 35 岁)、反复种植失败、复发性流产患者提高胚胎种植率、降低流产率、缩短妊娠时间的重要方法。常规 PGT-A 需要对胚胎进行有创活检, 可能会对胚胎后续发育造成潜在威胁及远期影响, 故寻找无创、安全、准确有效的胚胎染色体检测以及胚胎质量评估方法引起了生殖医学工作者的广泛关注。本文概述了近年来通过二代测序检测游离 DNA (cell-free DNA, cfDNA) 在无创胚胎植入前非整倍体检测 (non-invasive preimplantation genetic testing for aneuploidies, niPGT-A) 中的研究进展及其存在缺陷, 并提出应用单分子纳米孔技术检测 cfDNA 进行 niPGT-A 的创新性猜想, 为 niPGT-A 技术的进一步研究提供新的思路与参考。

【关键词】 无创胚胎植入前非整倍体检测; 游离 DNA; 单分子纳米孔 DNA 测序技术; 二代测序

Research progress and prospects of non-invasive preimplantation genetic testing technology

Huang Aomiao¹, Zhang Yinfeng², Liu Cong², Han Ying², Luo Haining²

¹ School of Medicine, Nankai University, Tianjin 300071, China; ² Reproductive Medicine Center, Tianjin Central Hospital of Obstetrics and Gynecology; Nankai University Affiliated Maternity Hospital, Tianjin 300100, China

Corresponding author: Luo Haining, Email: 30317012@nankai.edu.cn

【Abstract】 Preimplantation genetic testing for aneuploidy (PGT-A) is a crucial technique currently used to improve embryo implantation rate, reduce miscarriage rate, and shorten the time to pregnancy for patients who are of advanced maternal age (≥ 35 years), have experienced repeated implantation failures, or have a history of recurrent miscarriages. Conventional PGT-A necessitates an invasive embryo biopsy, which may pose potential risks to subsequent embryo development and long-term health outcomes. Consequently, there is a growing interest among reproductive medicine professionals in developing non-invasive, safe, accurate, and effective methods for assessing embryo chromosomal status and quality. This paper provides an overview of recent advancements in the use of next-generation sequencing for detecting cell-free DNA (cfDNA) in non-invasive preimplantation genetic testing for aneuploidy (niPGT-A). The review highlights both the potential of this approach and its existing limitations. Additionally, the paper proposes a novel hypothesis regarding the application of single-molecule nanopore technology for cfDNA detection in niPGT-A, offering new insights and serving as a reference for future research in this field.

【Key words】 Non-invasive preimplantation genetic testing for aneuploidies; Cell-free DNA; Single-molecule nanopore DNA sequencing technology; Next-generation sequencing

综述

DOI: 10.3760/cma.j.cn101441-20250228-00097

收稿日期 2025-02-28 本文编辑 李天琦

引用本文: 李柯婧, 李沛, 尹争, 等. 丙戊酸对癫痫男性生殖功能影响的研究进展[J]. 中华生殖与避孕杂志, 2025, 45(8): 851-855. DOI: 10.3760/cma.j.cn101441-20250228-00097.

丙戊酸对癫痫男性生殖功能影响的研究进展

李柯婧¹ 李沛² 尹争¹ 李西军¹ 李欣¹

¹青海大学研究生院, 西宁 810007; ²青海省人民医院神经内科, 西

宁 810007

通信作者: 李沛, Email: lp_0971@163.com

【摘要】 丙戊酸作为传统的广谱抗癫痫药物，长期使用会增加药物不良反应的风险，其中代谢综合征的发生最为常见，且常致男性不育。因此深入探讨癫痫、丙戊酸、代谢综合征与生殖的关系，发病机理以及危险因素、治疗措施，有助于药物不良反应的预防，对临床安全合理用药有重要意义。本文对癫痫男性在口服丙戊酸药物治疗后发生药物诱导的代谢综合征与不育情况进行总结，以期优化治疗，减少药物不良反应提供新的研究思路。

【关键词】 丙戊酸； 癫痫； 代谢综合征； 生殖； 男性

基金项目：中国抗癫痫协会癫痫科研基金（2020014B）；青海省人民医院院内课题（2023-qhsrmyy-22）

Progress in study of effect of valproic acid on male reproductive function in epileptic adults

Li Kejing¹, Li Pei², Yin Zheng¹, Li Xijun¹, Li Xin¹

¹ Graduate School, Qinghai University, Xining 810007, China; ² Department of Neurology, Qinghai Provincial People's Hospital, Xining 810007, China

Corresponding author: Li Pei, Email: lp_0971@163.com

【 Abstract 】 Valproic acid (VPA) is a traditional broad-spectrum antiepileptic drug, but long-term use of VPA will increase the risk of adverse drug reactions, with metabolic syndrome being the most common and often leading to infertility in men. Therefore, it is crucial to explore the relationship between epilepsy, VPA, metabolic syndrome and reproduction, the underlying pathogenesis, risk factors, and therapeutic measures. This exploration is essential for preventing adverse drug reactions and for guiding safe and rational use of drugs in clinical practice. This article summarizes the incidence of drug-induced metabolic syndrome and infertility in men with epilepsy who are treated with oral VPA, aiming to provide new research insights for optimizing treatment and reducing drug side effects.

【Key words】 Valproic acid; Epilepsy; Metabolic syndrome; Reproduction; Men

Fund program: Epilepsy Research Fund of the Chinese Antiepileptic Association (2020014B); Intramural Project of Qinghai Provincial People's Hospital (2023-qhsrmyy-22)

综述

DOI: 10.3760/cma.j.cn101441-20250206-00047

收稿日期 2025-02-06 本文编辑 李天琦

引用本文：王超莹，姚桂东. 哺乳动物原核形成和消失的分子机制研究进展[J]. 中华生殖与避孕杂志, 2025, 45(8): 856-860. DOI: 10.3760/cma.j.cn101441-20250206-00047.

哺乳动物原核形成和消失的分子机制研究进展

王超莹 姚桂东

郑州大学第一附属医院生殖医学中心, 郑州 450052

通信作者: 姚桂东, Email: ygdzzu@163.com

【摘要】 哺乳动物受精是一个高度复杂且受精精密调控的过程, 涉及精子与卵母细胞融合以及随后的细胞命运决定。本文对哺乳动物受精过程中原核形成和消失的分子机制进行综述, 特别关注原核形成及消失过程中的关键分子和调控机制, 并进一步探讨了原核形成和消失失败可能导致胚胎发育异常等引起不孕不育的问题, 以及解决原核形成或消失异常的方案, 旨在为生殖生物学和辅助生殖技术过程中导致原核形成和消失异常的分子机制提供理论依据, 并指出未来研究的潜在方向。

【关键词】 卵母细胞; 受精; 胚胎发育; 原核形成; 原核消失

基金项目: 国家自然科学基金 (U1904138)

Research advances in the molecular mechanisms underlying pronuclear formation and disappearance in mammals

Wang Chaoying, Yao Guidong

Center for Reproductive Medicine, the First Affiliated Hospital of Zhengzhou University, Zhengzhou 450052, China

Corresponding author: Yao Guidong, Email: ygdzzu@163.com

【 Abstract 】 Mammalian fertilization is a highly complex and finely regulated process that involves sperm-oocyte fusion and subsequent cell fate decisions. This article reviews the molecular mechanisms underlying pronucleus formation and disappearance during mammalian fertilization, with particular focus on the key molecules and regulatory mechanisms involved in these processes. It further discusses how failures in pronucleus formation and disappearance may lead to abnormal embryonic development and various infertility issues, along with potential solutions to address these abnormalities. The aim is to provide a theoretical foundation for understanding the molecular mechanisms that contribute to abnormal pronucleus formation and disappearance in the context of reproductive biology and assisted reproductive technology, while also highlighting potential directions for future research.

【 Key words 】 Oocytes; Fertilization; Embryonic development; Pronuclear formation; Pronuclear disappearance

Fund program: National Natural Science Foundation of China (U1904138)

经验交流

DOI: 10.3760/cma.j.cn101441-20250319-00128

收稿日期 2025-03-19 本文编辑 李天琦

引用本文：刘浏，朱勇，梁国庆，等. 人类精子库供精者遗传咨询的实践经验[J]. 中华生殖与避孕杂志, 2025, 45(8): 861-864. DOI: 10.3760/cma.j.cn101441-20250319-00128.

人类精子库供精者遗传咨询的实践经验

刘浏¹ 朱勇¹ 梁国庆¹ 朱宏¹ 江峰²

¹复旦大学附属妇产科医院人类精子库, 上海 200011; ²上海集爱遗传与不育诊疗中心男科, 上海 200011

通信作者：江峰, Email: jfsy001@163.com

【摘要】 基于复旦大学附属妇产科医院人类精子库的实践经验, 本文探讨供精者遗传咨询的必要性、核心内容、流程及常见问题, 并提出优化策略。供精者遗传咨询通过家族史调查、染色体核型分析、高通量基因测序及基因匹配等手段, 有效识别并降低供精子代的遗传风险, 提高精子库质量控制体系的科学性和可操作性。针对供精者遗传咨询面临的接受度问题、技术局限性、基因匹配复杂性、伦理问题、隐私保护及长期健康监测等问题, 本文提出优化基因筛查策略、建立智能化基因匹配系统、强化隐私保护机制及完善长期健康管理等应对措施, 以提高遗传咨询的精准性、安全性和可持续性。本文旨在为同行提供科学合理的实践参考, 助力优化精子库遗传咨询管理体系, 确保供精质量与辅助生殖的安全性。

【关键词】 精子库; 遗传咨询; 基因筛查; 基因匹配

Practical experience in genetic counseling for sperm donors in human sperm bank

Liu Liu¹, Zhu Yong¹, Liang Guoqing¹, Zhu Hong¹, Jiang Feng²

¹ Human Sperm Bank, Obstetrics and Gynecology Hospital, Fudan University, Shanghai 200011, China; ² Department of Andrology, Shanghai JIAI Genetics & IVF Institute-China USA Center, Shanghai 200011, China

Corresponding author: Jiang Feng, Email: jfsy001@163.com

【Abstract】 Based on the practical experience of the Human Sperm Bank, Obstetrics and Gynecology Hospital, Fudan University, this paper explores the necessity, core content, process, and common challenges of genetic counseling for sperm donors, while proposing optimization strategies. Genetic counseling for sperm donors employs methods such as family history investigation, karyotype analysis, high-throughput gene sequencing, and genetic matching to effectively identify and mitigate hereditary risks in donor-conceived offspring, thereby enhancing the scientific rigor and operability of sperm bank quality control systems. Addressing challenges such as donor acceptance, technological limitations, the complexity of genetic matching, ethical issues and privacy protection, and long-term health monitoring, this paper suggests strategies including optimizing

genetic screening protocols, establishing intelligent genetic matching systems, strengthening privacy protection mechanisms, and improving long-term health management. The aim of this paper is to provide a scientific and practical reference for professionals to optimize genetic counseling management in sperm banks, ensuring sperm donor quality and the safety of assisted reproduction.

【Key words】 Sperm bank; Genetic counseling; Genetic screening; Genetic matching