

· 综述 ·

孕激素受体膜元件1在颗粒细胞中的功能 及女性生殖中的作用

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【摘要】孕激素受体膜组件1(PGRMC1)是近年来新发现的特殊受体,属于膜相关的孕激素受体(MAPR)蛋白家族。PGRMC1介导孕酮(P₄)的抗有丝分裂和抗凋亡作用,并参与了颗粒细胞合成甾体激素的过程。在临床上,PGRMC1的异常表达与卵巢早衰(POF)及多囊卵巢综合征(PCOS)相关。本文就PGRMC1对颗粒细胞功能的影响、与卵泡发育的关系以及在人类生殖中的作用展开综述。

关键词:孕激素受体膜元件1(PGRMC1); 颗粒细胞(GC); 卵泡发育; 卵巢早衰(POF); 多囊卵巢综合征(PCOS)

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• 综述 •

Role of progesterone receptor membrane component 1 in granulosa cells function and female fertility

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【ABSTRACT】 Progesterone receptor membrane component 1 (PGRMC1), which belongs to the membrane-related progesterone receptor (MAPR) protein family, has been known as a novel special receptor in recent years. PGRMC1 mediated progesterone's action of anti-mitotic and anti-apoptotic. Moreover, PGRMC1 is also involved in steroid synthesis of granulosa cells. In the clinic, abnormal PGRMC1 expression has been proved to be related with premature ovarian failure (POF) and polycystic ovary syndrome (PCOS). The purpose of this article is to summarize the effect of PGRMC1 on granulosa cell function, the relationship between PGRMC1 and follicle development as well as the effect of PGRMC1 on human reproduction.

Key words: progesterone receptor membrane component 1 (PGRMC1); granulosa cells (GCs); follicle development; premature ovarian failure (POF); polycystic ovary syndrome (PCOS)

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粒细胞集落刺激因子对子宫内膜作用的研究

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【摘要】 子宫内膜容受性(ER)是胚胎着床的重要因素, 薄型子宫内膜(thin endometrium)是导致ER差的原因之一。薄型子宫内膜的发病机制、内膜厚度及血流灌注都与其胚胎移植的成功率息息相关, 因此薄型子宫内膜的治疗显得尤为重要。近年来的一些研究证实了粒细胞集落刺激因子(G-CSF)在子宫内膜修复方面起到了积极的促进作用, 探讨总结粒细胞集落刺激因子对子宫内膜的修复机制对胚胎移植适应症及临床应用具有重要意义。

关键词: 子宫内膜; 粒细胞集落刺激因子(G-CSF); 机制

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Recent advances of gene sequencing in preimplantation genetic diagnosis

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【ABSTRACT】 Gene sequencing technologies have undergone the development from the initial Sanger sequencing to modern single molecule sequencing since the last 30 years. Nowadays, Sanger sequencing has gradually been replaced by fluorescence *in situ* hybridization (FISH), comparative genomic hybridization (CGH), and microarray technology due to its low throughput and slow speed. As a new method for preimplantation genetic diagnosis (PGD), next-generation sequencing (NGS) not only can be applied to the detection of chromosomal aneuploidy, chromosomal structure abnormalities and single gene diseases, but also has higher precision and avoids the affection of probes. Mutated allele revealed by sequencing with aneuploidy and linkage analyse (MARSALA), a new technology based on NGS, has advantages for detecting chromosomal disorders and single gene disorders simultaneously. This review summarizes the developmental process of gene sequencing technologies and their applications in PGD, sums up the advantages and limitations of NGS technologies including MALBAC amplification technology and MARSALA technology which were built up recently when applied to NGS.

Key words: gene sequencing; next-generation sequencing (NGS); preimplantation genetic diagnosis (PGD);

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哺乳动物黄体生成素(LH)峰效应由颗粒细胞向 卵母细胞传递的分子通路

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【摘要】 血液中黄体生成素(luteinizing hormone, LH)在排卵前形成一个极大的峰值, LH与壁层颗粒细胞中的LH受体(LHR)结合诱导类表皮生长因子(EGF)的表达, 通过自分泌和旁分泌的作用激活颗粒细胞与卵丘细胞中的EGF受体(EGFR)、大鼠肉瘤病毒致癌基因(KRAS)、细胞外信号调节激酶1/2(ERK1/2), 激活的ERK1/2诱导表达前列腺素合成酶2(PTGS2)、类固醇合成快速调节蛋白(STAR)、透明质酸合成酶2(HAS2)、肿瘤坏死因子 α 诱导蛋白6(TNFAIP6)产生的前列腺素又和卵丘细胞上的PTGER2结合, 激活p38MAPK, 这一LH作用的信号转导通路最终刺激卵丘扩展卵母细胞成熟并最终排卵。

关键词: 排卵; 黄体生成素(LH); 分子机制; 细胞外信号调节激酶1/2(ERK1/2)

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Signals stimulated by LH transfer from granulosa cell to oocyte in mammalian follicle

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【ABSTRACT】 The level of luteinizing hormone (LH) in blood starts to rise and to form a peak before ovulation. LH binds to the receptor in mural granulosa cells to induce the expression of epidermal growth factor (EGF)-like factors, EGF receptor (EGFR), kirsten rat sarcoma viral oncogene homolog (*KRAS*), extracellular signal-regulated kinases 1 and 2 (ERK1/2), the activated ERK1/2 induces the expression of PTGS2, STAR, HAS2, TNFAIP6. Increased production of prostaglandins then provide ligands that bind PTGER2 on cumulus cells that activate p38MAPK. This program fulfills the role of autocrine and paracrine signals propagating the LH stimulus, leading to cumulus cell-oocyte complex expansion and oocyte maturation.

Key words: ovulation; luteinizing hormone (LH); molecular; extracellular signal-regulated kinases 1 and 2 (ERK1/2)

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人胎盘滋养细胞分选技术研究进展

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【摘要】人胎盘滋养细胞(TB)包括绒毛细胞滋养细胞(VCT)、合体滋养细胞(VST)和绒毛外细胞滋养细胞(EVT)。各种TB形态差异较大,在母-胎界面的功能也各不相同,此外由于母体细胞的污染,因此有必要对TB的类别加以区分。如何在保证细胞活性的同时提高纯度成为构建TB细胞系的主要困难之一。梯度离心、磁珠分选和流式细胞术是广泛应用的细胞分选技术。各种分选技术在TB研究中各有利弊,磁珠分选结合流式细胞术是目前较为有效的分选策略。

关键词: 滋养细胞(TB); 流式细胞术; 磁珠分选

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Research progress of human placental trophoblasts sorting skills

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【ABSTRACT】 Human placental trophoblast cell (TB) can be classified into villous cytotrophoblast cell (VCT), villous syncytiotrophoblast cell (VST), and extravillous trophoblast cell (EVT). Varieties of TB are differential in morphology, and play disparate roles in the physiological or pathological processes. Besides inevitable maternal cell contamination, it is necessary to highlight the purity of TB in researches, in which also exists the contradiction of vitality and purity problems. Density gradient centrifugation, immunomagnetic separation and flow cytometry (FCM) have been used widely in the sorting of cells. There are advantages and disadvantages for all of these methods, however, immunomagnetic separation combined with FCM appears to be more effective among all the sorting tactics.

Key words: trophoblasts (TB); flow cytometry (FCM); immunomagnetic separation

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自噬机制与卵巢早衰的关系

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【摘要】细胞自噬是哺乳动物细胞物质代谢的一种重要机制,细胞自噬异常将导致细胞功能异常甚至死亡。细胞自噬异常在卵巢早衰(POF)的发病中扮演着重要的角色,细胞自噬与细胞凋亡之间存在着许多分子联系,凋亡相关蛋白 Bcl-2、Caspase 与自噬相关蛋白 Beclin-1 相互作用,可以引起卵巢发育异常;凋亡蛋白 Bim、Fas 与自噬相关信号通路 PTEN-PI3K 及下游转录因子 FoxO 蛋白相互作用,可以导致卵巢卵泡激活异常;激素异常通过 TGF- β /Smad3 通路介导凋亡和自噬相关蛋白异常,导致卵泡闭锁,从而可以导致 POF 的发病。细胞自噬极有可能与细胞凋亡一起,是 POF 的主要发病机制。

关键词: 自噬; 卵巢早衰(POF); 卵泡

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Relationship between autophagy mechanism and premature ovarian failure

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【ABSTRACT】 Autophagy is an important mechanism of metabolism of mammalian cells. Abnormal cell autophagy will lead to abnormal cell function and even death. Abnormal cell autophagy plays an important role in the pathogenesis of premature ovarian failure (POF). There are many molecular links between autophagy and apoptosis. The interaction among apoptosis related proteins Bcl-2, caspase-3 and autophagy associated protein Beclin-1 can cause abnormal ovarian development; and the interaction among apoptosis protein BIM, Fas and autophagy related pathways PTEN-PI3K and downstream transcription factor FOXO protein can lead to abnormal activation of ovarian follicle; those abnormal hormone levels through the TGF-beta/Smad3 pathway mediates apoptosis and autophagy associated protein abnormalities lead to follicular atresia, and can lead to the further occurrence of POF. The main pathogenesis of POF may be the result of the interaction between autophagy and apoptosis.

Key words: autophagy; premature ovarian failure (POF); follicle

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卵巢早衰的遗传学研究进展

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【摘要】 卵巢早衰(POF)是一种临床高度异质且病因复杂的疾病, 遗传因素是其重要的发病原因。遗传因素主要包括 X 染色体、常染色体异常、微小 RNA 表达异常等。全基因组关联研究(GWAS)、高通量测序(NGS)技术等迅猛发展, 可对基因标志物或核苷酸多态性进行统计分析, 发现其间的内部联系, 揭示新的致病基因, 为 POF 的遗传学基础研究提供崭新的平台。

关键词: 卵巢早衰(POF); 遗传学; 病因学

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Genetics research progress of premature ovarian failure

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【ABSTRACT】 Premature ovarian failure (POF) is a heterogeneous disease and its etiology is complicated. Genetic factor is the most common cause of POF. The genetic abnormalities of X-chromosome, candidate genes and microRNAs all play important roles in POF pathogenesis. The unbiased approaches of genome-wide association studies (GWAS) and next-generation sequencing (NGS) technologies have provided a greater insight into POF pathogenesis. In this review, we provide the progress of the genetic basis of POF pathogenesis.

Key words: premature ovarian failure (POF); genetics; etiology

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抗苗勒氏管激素评估卵巢储备功能的临床应用现状

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【摘要】 卵巢储备功能指卵巢内存留卵泡的数量和质量, 其能间接地反映女性的生育能力。抗苗勒氏管激素(anti-Müllerian hormone, AMH)作为卵巢特异性的生长因子, 由颗粒细胞分泌, 是预测卵巢储备功能的一个有效指标。血清AMH的水平可以预测卵泡池的大小、卵巢的反应性及女性的绝经年龄, 同时可以评估癌症患者的卵巢储备功能。但AMH的测定值受到多种因素的影响, 存在一定的局限性。

关键词: 抗苗勒氏管激素(AMH); 卵巢储备; 生育能力; 局限性

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Clinical status of anti-Müllerian hormone (AMH) in the evaluation of ovarian reserve function

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【ABSTRACT】 Ovarian reserve refers to the quantity and quality of remaining follicles in ovaries, which can indirectly reflect the fertility of the female. Anti-Müllerian hormone (AMH) as the ovary-specific growth factor secreted by granulosa cells, is currently an effective indicator for predicting ovarian reserve function. The serum AMH level can predict the size of the follicle pool, ovarian response and the menopause age, but also can evaluate the ovarian function in patients with cancer. But the measurement of AMH level is influenced by many factors, thus the application of AMH still has some limitations.

Key words: anti-Müllerian hormone (AMH); ovarian reserve; fertility; limitation

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抗苗勒氏管激素与卵巢储备及反应性 相关研究进展

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【摘要】抗苗勒氏管激素(AMH)的水平对卵巢中的卵泡发育与类固醇激素的产生具有重要意义。体循环中AMH水平受性别和年龄等因素影响,在性腺发育和生殖系统疾病的评价中至关重要。AMH可预测卵巢在辅助生殖技术(assisted reproductive technology, ART)中的反应性及女性绝经时间等,并反映卵巢手术或损伤后的卵巢储备能力。本文对近年来关于AMH评价卵巢储备及ART中卵巢反应性的相关研究成果进行综述。

关键词: 抗苗勒氏管激素(AMH); 卵巢储备; 卵巢反应性

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Research advances of the relationship between anti-Müllerian hormone with ovarian reserve and response

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【ABSTRACT】 The level of anti-Müllerian hormone (AMH) was important for the development of ovarian follicles and the production of steroid hormones. The level of AMH in body circulation was influenced by gender and age, which was vital to evaluate gonadal developments and reproductive system diseases. AMH not only could predict ovarian response during assisted reproductive technology (ART), but also could predict the duration of menopause, and also reflected the ovarian reserve capacity after ovarian surgery or injury. This article summarized recent studies on the evaluation of ovarian reserve and ovarian response in ART with AMH, in order to provide reference for clinical diagnosis and treatment of female reproductive system diseases.

Key words: anti-Müllerian hormone (AMH); ovarian reserve; ovarian response

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内分泌腺来源的血管内皮生长因子 在女性生殖中的研究进展

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【摘要】 内分泌腺来源的血管内皮生长因子(EG-VEGF)是近年研究发现的一种新的血管因子, 其结构与VEGF不同而功能类似。作为迄今被发现的唯一的组织特异性血管生成因子, EG-VEGF在女性生殖系统中较VEGF有更为特异的功能, EG-VEGF在女性的月经周期、胚胎着床、妊娠维持等生殖功能中起重要作用, 其功能亢进或不足与多种病理状态相关, 但其作用机制尚未完全阐明。

关键词: 内分泌腺来源的血管内皮生长因子(EG-VEGF); 生殖生理; 生殖病理; 血管发育

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Research progress of endocrine gland-derived vascular endothelial growth factor in female reproduction

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【ABSTRACT】 Endocrine gland-derived vascular endothelial growth factor (EG-VEGF) is a new vascular factor, which has been discovered in recent years with different structure but similar function compared with VEGF. As the only tissue-specific angiogenic factor found so far, the function of EG-VEGF in female reproductive system is more specific than that of VEGF. EG-VEGF plays an important role in female reproductive endocrine system, including menstrual cycle, embryo implantation and pregnancy maintenance etc. Its hyperfunction or insufficiency is associated with a variety of pathological conditions. However, its mechanism has not been well elucidated.

Key words: endocrine gland-derived vascular endothelial growth factor (EG-VEGF); reproductive physiology; reproductive pathology; vascular development

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基因测序在胚胎植入前遗传学诊断应用的研究进展

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【摘要】 经过30多年的发展, 基因测序技术已从最初的Sanger测序发展至当今以单分子测序为特点的测序。早期的Sanger测序可应用于单基因疾病的检测, 但其可检测的通量小、速度慢, 逐渐被荧光原位杂交(FISH)、比较基因组杂交(CGH)、芯片检测技术等取代。下一代测序(next-generation sequencing, NGS)技术作为胚胎植入前遗传学诊断(preimplantation genetic diagnosis, PGD)的新检测手段, 不仅能检测染色体非整倍性、染色体结构异常以及单基因疾病, 而且精度更高, 弥补了芯片检测易受探针影响的缺陷。新近建立的基于NGS的非整倍体测序与连锁分析(mutated allele revealed by sequencing with aneuploidy and linkage analyses, MARSALA)技术可以同时检测染色体疾病和单基因疾病。本文概述了基因测序技术的发展进程及其在PGD中的应用, 介绍了包括近年开发的多重退火环状循环扩增(MALBAC)技术和MARSALA在内的NGS技术应用用于PGD的优点和局限。

关键词: 基因测序; 下一代测序(NGS); 胚胎植入前遗传学诊断(PGD); 辅助生殖技术

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Recent advances of gene sequencing in preimplantation genetic diagnosis

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【ABSTRACT】 Gene sequencing technologies have undergone the development from the initial Sanger sequencing to modern single molecule sequencing since the last 30 years. Nowadays, Sanger sequencing has gradually been replaced by fluorescence *in situ* hybridization (FISH), comparative genomic hybridization (CGH), and microarray technology due to its low throughput and slow speed. As a new method for preimplantation genetic diagnosis (PGD), next-generation sequencing (NGS) not only can be applied to the detection of chromosomal aneuploidy, chromosomal structure abnormalities and single gene diseases, but also has higher precision and avoids the affection of probes. Mutated allele revealed by sequencing with aneuploidy and linkage analysis (MARSALA), a new technology based on NGS, has advantages for detecting chromosomal disorders and single gene disorders simultaneously. This review summarizes the developmental process of gene sequencing technologies and their applications in PGD, sums up the advantages and limitations of NGS technologies including MALBAC amplification technology and MARSALA technology which were built up recently when applied to NGS.

Key words: gene sequencing; next-generation sequencing (NGS); preimplantation genetic diagnosis (PGD); assisted reproductive technology

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HOXA10基因与不孕症关系的研究进展

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【摘要】 子宫内膜容受性(ER)的建立是妊娠的关键性前提。ER的降低是不孕症的主要因素之一。HOXA10基因近年来被认为在ER的建立过程中具有重要意义, 子宫内膜HOXA10表达增加是内膜具有良好容受性的必要条件, 与胚胎着床密切相关, 目前已将HOXA10作为ER建立的标志物之一。本文旨在对于子宫内膜异位症(EMS)、多囊卵巢综合征(PCOS)、输卵管积水及控制性超促排卵(COH)治疗不孕症患者的HOXA10基因是否存在异常表达的相关研究进展进行综述, 希望能为治疗相关不孕症提供新的思路。

关键词: HOXA10 基因; 不孕症; 子宫内膜容受性(ER)

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Research progress of the relationship between *HOXA10* gene and infertility

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【ABSTRACT】 The establishment of endometrial receptivity (ER) is a crucial prerequisite for pregnancy. Insufficient of ER is one of the main factors of infertility. In recent years, *HOXA10* gene was found to have an important significance in the process of establishing ER, and the increase in endometrial *HOXA10* expression is necessary for well ER, which is closely related to embryo implantation. Nowadays, *HOXA10* has been treated as one marker of ER. This article is intended to review the relevant research progress about the expression of *HOXA10* gene in the endometriosis (EMS), polycystic ovary syndrome (PCOS), hydrosalpinx, and the infertile patients treated by controlled ovarian hyperstimulation (COH), which is hoped to provide a new idea for the treatment of related infertility.

Key words: *HOXA10* gene; infertility; endometrial receptivity (ER)

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转基因治疗不孕相关疾病研究进展

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【摘要】转基因治疗可以在一定程度上纠正或补偿基因异常或缺陷所导致的不孕相关疾病, 以使部分患者获得生育能力。但现阶段转基因治疗仍处于初始阶段, 应用范围有限, 并且其体内研究均使用的动物模型, 因涉及到伦理及潜在的安全问题限制了其在人体的直接应用。本综述分别从女性不孕、男性不育两方面对转基因治疗的研究进展进行总结, 为今后不孕相关疾病的治疗提供了一个新的思路。

关键词: 不育; 基因异常; 转基因治疗

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Research progress of gene transfer therapy for infertility

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【ABSTRACT】 Gene transfer therapy can treat some infertile diseases by correcting or compensating genetic abnormalities or defects to some extent. However, gene transfer therapy is still in the initial stage at present. Because of the ethics and potential safety problems involved, gene transfer therapy has not been widely applied, especially directly applied to the human body. All of the researches *in vivo* used animal models. The current review summarizes the research progress of gene transfer therapy in female and male infertility respectively, and provides a new thought for the future of infertility treatment.

Key words: infertility; genetic abnormality; gene transfer therapy

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孤雌胚胎干细胞在细胞疗法中的优势与局限性

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【摘要】孤雌胚胎干细胞(pESCs)是建立患者特异性多能干细胞的有效途径, 因具有类似胚胎干细胞(ESCs)自我复制和多潜能特性, 成为再生医学的“种子细胞”。杂合型pESCs有望用于自体移植, 纯合型pESCs理论上可用于异体基因疗法, 却因自然杀伤(NK)细胞识别而产生免疫排斥反应。本文旨在综述孤雌生殖的表观遗传特性, 总结pESCs在细胞疗法中的优势和局限性, 阐述克服免疫排斥反应的新策略。

关键词: 孤雌胚胎干细胞(pESCs); 基因组印记; 细胞疗法; 纯合型; 杂合型

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Advantages and limitations of parthenogenetic embryonic stem cells in cell therapy

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【ABSTRACT】 Parthenogenetic embryonic stem cells (pESCs), as “seed cells” for regenerative medicine, are an effective way to build specific pluripotent stem cells for patients, because of its similarity of self-renewal and pluripotent characteristics to embryonic stem cells (ESCs). Heterozygous pESCs is expected to be used for autologous transplantation, while homozygous pESCs can be used for allogeneic gene therapy in theory. In the paper, we review parthenogenesis epigenetic characteristics, and summarize the advantages and limitations of pESCs in the area of cell therapy, and elaborate the latest strategies of overcoming immune rejection.

Key words: parthenogenetic embryonic stem cells (pESCs); imprinted gene; cell therapy; homozygous; heterozygous

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miR-518b与不良妊娠结局的研究

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【摘要】 微小RNA(miRNA)通过与对应的靶基因结合, 调控生物体的生殖、发育等生命活动。miR-518b是一种胎盘特异性miRNA, 与子痫前期、完全性葡萄胎等不良妊娠结局发病有着密切的关系。miR-518b可以在孕妇外周血中检测出, 血清的检测可以为不良妊娠结局的诊断、监测与预防提供了新的方法, miR-518b的相关研究为不良妊娠结局的发病机理提供了新的研究思路。

关键词: 微小RNA(miRNA); miR-518b; 妊娠结局

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Researches of microRNA-518b and adverse pregnancy outcomes

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【ABSTRACT】 MicroRNAs (miRNAs) regulate reproductive development by combination with target genes. MiR-518b is placenta-specific, and closely related to adverse pregnancy outcomes, such as preeclampsia and complete hydatidiform moles. MiR-518b can be detected in peripheral blood of the pregnant women, which provides a new way to diagnose and prevents adverse pregnancy outcomes. The research of miR-518b provided a new idea to the pathogenesis of adverse pregnancy outcomes.

Key words: microRNA (miRNA); miR-518b; pregnancy outcome

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卵泡液代谢组学最新研究进展

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【摘要】代谢组学是继基因组学、转录组学、蛋白质组学之后系统生物学的主要研究平台,并逐渐成为生命科学的研究热点。在辅助生殖技术(assisted reproductive technology, ART)领域,卵泡液代谢组学已取得较大进展。目前,国内外大多数回顾性研究表明卵泡液相关代谢物(糖类、脂肪酸、氨基酸、激素等)可预测卵母细胞发育潜能,但有待大样本、多中心随机对照试验进一步证实。卵泡液代谢组学的深入研究有望成为评估卵母细胞质量并改善ART治疗结局的重要方法和手段。

关键词: 卵泡液(FF); 代谢组学; 卵母细胞; 胚胎

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Recent research progress of the follicular fluid metabonomics

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【ABSTRACT】 Following the genomics, transcriptomics and proteomics, metabonomics is becoming a focus of systems biology and a hotspot of life science research. In assisted reproductive technology (ART), follicular fluid (FF) metabonomics has obtained great progress. At present, most of the retrospective studies have found FF related metabolites (sugars, fatty acids, amino acids, hormones, etc.) can predict oocyte development potential, but clinical research needs to be further confirmed by using multi-center and randomized observation with expanding samples. In-depth study of FF metabonomics is expected to become the important approach and method of evaluating the oocyte quality and improving the treatment outcome of the ART.

Key words: follicular fluid (FF); metabolome; oocyte; embryo

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