

Applied Molecular Biology and Genetics in Obstetrics and Gynecology

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Abstract

Introduction: Molecular biology and genetics now occupy a central place in obstetrics and gynecology, offering innovative diagnostic, prognostic, and therapeutic tools. **Objective:** To describe the role of molecular biology and genetics in obstetrics and gynecology. **Applications in Gynecology:** In oncology, genetics allows us to identify new genes involved in breast, cervical, endometrial, and ovarian cancers, to identify high-risk patients, and to develop more tailored treatments to provide optimal care. Molecular biology and molecular genetics are essential for understanding infertility. They allow us to identify chromosomal, gene, and epigenetic abnormalities responsible for reproductive disorders. **Applications in Obstetrics:** Pregnancy is a complex physiological process involving genetic, epigenetic, hormonal, and immune interactions. Molecular abnormalities can disrupt its proper course, leading to various maternal or fetal pathologies. Prenatal diagnosis increasingly relies on molecular biology and molecular genetics. **Conclusion:** molecular biology and genetics allow for a better understanding of the fundamental mechanisms of reproduction, explain many causes of infertility, and shed light on the pathophysiology of the main pregnancy pathologies. These advances constitute not only powerful diagnostic and prognostic tools, but also drivers of progress toward precision obstetrics and gynecology, serving the health of women and the mother-child dyad.

Keywords

Biology, Genetics, Cancer, Infertility, Pregnancy, Burkina Faso

1. Introduction

Molecular biology involves integrating all the molecular data necessary for under-

standing biological mechanisms. The term “molecular biology,” commonly used in medical biology, refers to the study of nucleic acids, specifically deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). DNA carries genetic information. Its alterations can cause congenital or acquired diseases. The advent of molecular biology has revolutionized biology, particularly genetics, and has led, among other things, to a better understanding of the pathophysiological mechanisms of genetically transmitted diseases as well as certain acquired conditions [1]-[3]. Obstetrics and gynecology, like other medical disciplines, has greatly benefited from these increasingly powerful techniques. This chapter will address molecular biology and genetics as applied to obstetrics and gynecology.

2. Basic Reminders

❖ Foundations of biology and molecular genetics

All cells store their hereditary information in DNA. Eukaryotic DNA is packaged into a set of chromosomes (46 chromosomes, including 44 autosomes and one pair of sex chromosomes, XX in females; XY in males). Cells replicate their hereditary information by template-based polymerization. They transcribe parts of their hereditary information into RNA. The RNA is then translated into protein by ribosomes. The gene is expressed as a protein. The gene represents the fragment of genetic information corresponding to a protein [2] [4].

❖ Specificity in obstetrics and gynecology

Obstetrics and gynecology is a medical discipline that deals with women at all stages of life (from puberty to menopause and beyond), including pregnancy monitoring, childbirth, postpartum care, and the prevention, diagnosis, and treatment of diseases of the female reproductive system and breasts. Obstetrics and gynecology is distinguished by its unique specialization in molecular biology and genetics. It involves two patients simultaneously: the mother and the unborn child, and sometimes the biological father, making the applications of molecular biology and genetics particularly strategic [5].

3. Applications in Gynecology

❖ Diagnosis and treatment of gynecological cancers

Cancer is a complex group of diseases resulting from genetic and epigenetic alterations that interfere with cell growth and death [6]. In oncology, genetics allows us to identify new genes involved in breast, cervical, endometrial and ovarian cancers, to identify high-risk patients and to develop more suitable treatments in order to offer optimal care. The involvement of various molecules in the oncogenic processes of invasion, metastasis, and treatment resistance has been demonstrated. The exponential growth of genomic and proteomic data collected over the last twenty years has led to major advances in understanding the molecular mechanisms of gynecological cancers, which have been applied to diagnostic and therapeutic strategies. Targeted therapies have been developed and adopted, particularly for advanced, refractory, or recurrent cancers, based on individual molecular

profiles [6]-[10].

❖ Infertility and medically assisted reproduction

Infertility is a major human reproductive health problem, affecting approximately 15% of couples worldwide [11]. It is estimated that half of infertility cases have a genetic component. Molecular biology and molecular genetics are essential for understanding infertility. The advent of high-throughput sequencing technologies has greatly facilitated the identification of genetic mutations associated with infertility in patients over the past 20 years. They allow the identification of chromosomal, gene, and epigenetic abnormalities responsible for reproductive disorders, guide diagnosis, personalize care, and pave the way for innovative therapeutic approaches. In female infertility, chromosomal abnormalities (Turner syndrome) and genetic mutations responsible for early menopause have been identified through molecular biology, as well as microdeletions of the Y chromosome in azoospermia or severe oligospermia, with emerging therapeutic possibilities such as genome editing (CRISPR-Cas9) and regenerative medicine (stem cells for gametogenesis) [12]-[16].

In assisted reproductive technology (ART), preimplantation genetic diagnosis (PGD) allows for the assessment of the risks of the occurrence or recurrence of a genetic disorder. In the context of egg donation, genetic screening is a current issue that raises numerous ethical questions related to the improvement of techniques enabling the detection of an increasing number of genome mutations [17] [18].

4. Applications in Obstetrics

❖ Pregnancy pathologies

Pregnancy is a complex physiological process involving genetic, epigenetic, hormonal, and immune interactions. Molecular abnormalities can disrupt its proper course, leading to various maternal or fetal pathologies. These main pathologies include preeclampsia and its complications, and recurrent miscarriages related to miRNA expression, gestational diabetes, threatened preterm labor, hydatidiform mole, Beckwith-Wiedemann syndrome (11p15 methylation alterations) and Silver-Russell syndrome (hypomethylation locus 11p15, chromosome 7 abnormalities) [19]-[30].

❖ Prenatal diagnosis

Antenatal diagnosis encompasses all examinations performed during pregnancy to detect chromosomal, genetic, metabolic, or morphological abnormalities in the embryo or fetus and to enable early intervention (medical, surgical, therapeutic, or even decision-making). Antenatal diagnosis increasingly relies on molecular biology and molecular genetics. These technologies offer increasingly precise and often non-invasive tools, ranging from non-invasive prenatal screening (circulating fetal DNA) to next-generation sequencing (NGS). They allow us to go beyond morphological screening to identify chromosomal (trisomy), monogenic (sickle cell disease, hemophilia), or epigenetic abnormalities, thus enabling preci-

sion fetal medicine [31] [32].

5. Conclusion

Molecular biology and genetics now occupy a central place in obstetrics and gynecology. They allow for a better understanding of the fundamental mechanisms of reproduction, explain many causes of infertility, and shed light on the pathophysiology of the main pregnancy complications. These advances constitute not only powerful diagnostic and prognostic tools, but also drivers of progress toward precision obstetrics and gynecology, serving the health of women and the mother-child dyad.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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