

Literature Review

Progress, Consensus and Controversies in Gynecological and Obstetric Screening: A Narrative Literature Review

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Abstract

Screening in gynecology and obstetrics has been transformed by molecular biology, non-invasive prenatal diagnosis, and advanced imaging. This narrative literature review analyzes current scientific evidence supporting primary and secondary prevention strategies. In gynecology, we examine the validated superiority of the HPV DNA test over cervical cytology as well as the role of digital breast tomosynthesis in breast screening. In obstetrics, we detail early prediction of pre-eclampsia using biophysical and angiogenic markers, the revolution of NIPT for aneuploidies, and the management of gestational diabetes. Finally, ethical issues, equitable access, overdiagnosis, and the emerging integration of artificial intelligence are discussed.

Introduction

Preventive medicine in women's health relies on the early identification of potentially serious conditions in asymptomatic individuals. The historical criteria of Wilson and Jungner remain the foundation for evaluating any screening program: the disease must constitute a major public health problem, its natural history must be understood, and an effective treatment must be available at a pre-clinical stage [1].

In obstetrics and gynecology, the screening paradigm has shifted over the past decade. The traditional one-size-fits-all approach is increasingly being replaced by personalized risk stratification. The aim of this narrative review is to synthesize current evidence, assess international consensus, and identify future clinical perspectives based on major guidelines and clinical studies published over recent years.

Documentary research strategy

A comprehensive search was conducted across the PubMed/MEDLINE, Embase, and Cochrane Library databases for works published between January 2018 and early 2026. The literature search targeted broad aspects of preventive screening in obstetrics and gynecology.

MeSH descriptors & keywords: "Mass Screening",

"Uterine Cervical Neoplasms", "HPV DNA Tests", "Breast Neoplasms", "Prenatal Diagnosis", "Non-Invasive Prenatal Testing (NIPT)", "Pre-Eclampsia/prevention and control", "Pregnancy in Diabetics".

Selection criteria: Recommendations and consensus guidelines from international learned societies (FIGO, ACOG, RCOG, CNGOF, WHO).

Meta-analyses, systematic reviews, and major randomized controlled trials (RCTs).

Initial electronic searches yielded over 320 records; after removing duplicates and screening titles/abstracts, 24 pivotal articles and clinical recommendations were retained for synthesis in this review.

Exclusion criteria: Isolated case studies, case series with fewer than 50 patients, articles not peer-reviewed, and non-English/non-French language publications without available translation.

Screening in gynecology

Cervical cancer: The transition to molecular biology

The traditional Pap smear has largely given way to

More Information

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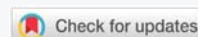
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molecular tests for detecting high-risk Human Papillomavirus DNA (HR-HPV) [2,3].

Clinical superiority: Meta-analyses confirm that primary HPV testing exhibits a sensitivity exceeding 95% for detecting grade 2 or higher cervical intraepithelial neoplasia (CIN2+), compared to approximately 50–60% for cytology alone [4]. The very high negative predictive value (NPV) of HPV testing allows for a safe extension of the screening interval to 5 years in screen-negative women.

Triage algorithms: Given the high prevalence of transient HPV infections, a positive test requires systematic triage (HPV 16/18 genotyping and/or reflex cytology) to avoid unnecessary colposcopies and over-management [5].

Accessibility: Self-sampling of vaginal specimens has demonstrated diagnostic performance equivalent to clinician-collected samples, representing a key strategy to reach underserved populations not participating in organized screening [6].

Breast cancer: risk stratification and digital imaging

Biennial mammographic screening remains the global standard for average-risk women aged 50 to 74 [7]. However, limitations of standard 2D digital mammography in women with dense breasts (BI-RADS C and D) have driven technological advancements:

3D Digital Breast Tomosynthesis (DBT): Increases the detection rate of invasive cancers while significantly reducing recall rates for false positives compared to 2D mammography alone [8].

Breast Ultrasound and MRI: Supplemental MRI is recommended in women at high familial or genetic risk (e.g., BRCA1/2 or PALB2 pathogenic variants) [9]. Ultrasound remains a widely used adjunct for dense breast parenchyma when MRI is unavailable.

Screening in obstetrics and fetal medicine

The operational window for obstetric screening and early intervention has shifted predominantly from the third to the first trimester of pregnancy through combined clinical, ultrasound, and biochemical risk predictive models ([10].

Discussion: Ethical Issues, limitations, and future prospects

Risks of overdiagnosis and overtreatment

The increased sensitivity of modern diagnostic tools inevitably carries the risk of identifying transient or non-progressive lesions. In cervical screening as well as in breast imaging, avoiding overtreatment of indolent, asymptomatic lesions remains a major public health challenge.

Contribution of Artificial Intelligence (AI)

The integration of deep learning algorithms is accelerating diagnostic workflow efficiency:

In breast imaging: Automated triage of normal mammograms to decrease workload for second readers and reduce human fatigue errors.

In fetal ultrasound: Automated recognition of standard anatomic planes and automated standardization of nuchal translucency and fetal biometric measurements.

Equity in public health

Despite technical advancements, access to screening programs remains unequal. Socio-economic barriers and regions suffering from healthcare worker shortages or inadequate medical infrastructure (medically underserved areas) necessitate the implementation of mobile screening units, tele-expertise, and point-of-care (POC) testing devices.

Conclusion

Modern screening in gynecology and obstetrics is rapidly progressing toward personalized, risk-stratified, and predictive care. Updating clinical workflows with robust evidence from the literature—while ensuring equitable access, avoiding overtreatment, and standardizing preventive dosages—is key to maximizing patient benefit and optimizing health system outcomes.

Regulatory declarations

Conflicts of interest: The author declares no conflicts of interest related to this article.

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Targeted Pathology	Screening Tools & Markers	Evaluation Window	Clinical Impact & Management
Down Syndrome & Common Aneuploidies	Combined 1st trimester screening (nuchal translucency, free β-hCG, PAPP-A) followed by NIPT (cell-free fetal DNA).	11+0 to 13+6 weeks of gestation (GW)	NIPT sensitivity > 99% for Trisomy 21 [11]. Achieves >90% reduction in invasive diagnostic procedures (amniocentesis).
Early Pre-eclampsia	Combined risk algorithm: Mean arterial pressure, uterine artery Doppler, PlGF, and PAPP-A.	11+0 to 13+6 GW	Identification of high-risk women for preventive introduction of Aspirin (≥ 150 mg/day) initiated before 16 weeks of gestation [12,13]. Reduces preterm pre-eclampsia (<37 GW) by >60%.
Gestational Diabetes Mellitus (GDM)	1st trimester: Fasting plasma glucose in women with risk factors. 24–28 GW: 75g OGTT (IADPSG criteria).	1st trimester or 24–28 GW	Significant reduction in perinatal complications (macrosomia, shoulder dystocia, pre-eclampsia) through early lifestyle, dietary intervention, or pharmacotherapy [14].



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