PIPADOR REVISION

MRCOG Part 1 Revision Notes

Omics Technologies

BIOCHEMISTRY • LECTURE 237

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG PART 1 REVISION NOTES | BIOCHEMISTRY

237 Omics Technologies

Genomics, epigenomics, transcriptomics, proteomics, metabolomics and microbiomics — methods, interpretation and O&G applications

≈ 20,000	Mass spec	cfDNA	Multi-omics
Protein-coding genes	Core proteomics/ metabolomics tool	Basis of NIPT (≈ 10% fetal fraction)	Integrated systems biology

■ KEY FACTS: Learning objectives

- Define the main 'omics' technologies (genomics, epigenomics, transcriptomics, proteomics, metabolomics, microbiomics).
- Describe the principal methods (sequencing, microarrays, mass spectrometry, NMR) and bioinformatics.
- Describe current and emerging applications in O&G and their limitations.

237.1 The omics cascade

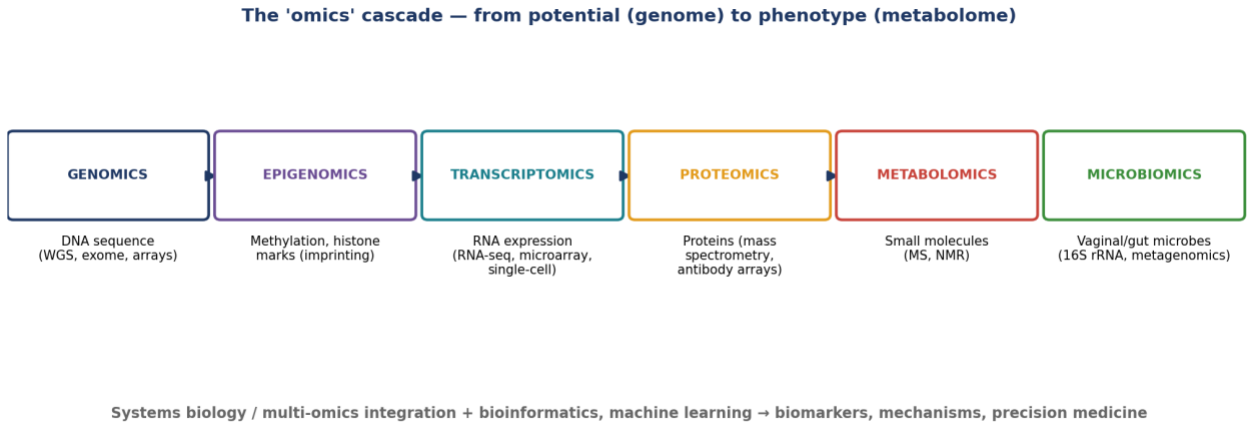


Figure 237.1 — From genome to metabolome

Omics	Studies	Key methods
Genomics	Entire DNA sequence and variation (SNPs, CNVs, mutations)	Next-generation sequencing (whole genome/exome), chromosomal microarray (CNVs), PCR, Sanger
Epigenomics	Heritable changes in gene expression without sequence change — DNA methylation, histone modifications, non-coding RNA	Bisulphite sequencing, ChIP-seq; imprinting (Beckwith–Wiedemann, Prader–Willi/Angelman), fetal programming
Transcriptomics	All RNA transcripts (expression)	RNA-seq, microarrays, single-cell RNA-seq (e.g. placental cell atlases)
Proteomics	All proteins, PTMs, interactions	Mass spectrometry (LC-MS/MS, MALDI-TOF), 2-D gels, antibody/aptamer arrays
Metabolomics	Small molecules (< 1.5 kDa) — the closest to phenotype	Mass spectrometry, NMR; newborn screening by tandem MS is a targeted metabolomic test
Microbiomics	Microbial communities	16S rRNA sequencing, shotgun metagenomics — vaginal microbiome (Lactobacillus-dominant vs diverse — preterm birth, BV)

Omics	Studies	Key methods
Lipidomics, glycomics	Lipid and glycan profiles	MS

237.2 Bioinformatics and interpretation

- Huge datasets need **bioinformatics** (alignment, variant calling, pathway analysis), **statistics** for multiple testing (false discovery rate) and increasingly **machine learning**.
- Variants classified by ACMG criteria (pathogenic → benign); **variants of uncertain significance (VUS)** are common — counselling and data-sharing essential.
- Limitations: cost, standardisation, sample handling (pre-analytical variation in proteins/metabolites), validation in independent cohorts, ethical issues (incidental findings, consent, data privacy, insurance).

237.3 Applications in O&G

Area	Omics application
Prenatal screening/diagnosis	NIPT — sequencing of cell-free fetal DNA (from placenta) for trisomies 21/18/13, fetal sex, fetal RhD genotyping, monogenic conditions; chromosomal microarray after abnormal scan; prenatal exome sequencing for structural anomalies (NHS Genomic Test Directory)
Pre-eclampsia	Proteomics/transcriptomics identified sFlt-1 and PlGF → now used clinically (NICE DG49); cfRNA signatures under research
Preterm birth	Cervicovaginal proteomics (fetal fibronectin, IGFBP-1), vaginal microbiome profiles, maternal cfRNA, metabolomic signatures
Gynaecological oncology	BRCA1/2 and HRD testing (PARP inhibitors), Lynch (MMR immunohistochemistry/MSI), molecular classification of endometrial cancer (POLE, MMRd, p53abn, NSMP — ProMisE/TCGA), ovarian cancer biomarkers (CA-125, HE4)
Reproductive medicine	Preimplantation genetic testing (PGT-A, PGT-M), endometrial receptivity transcriptomics (limited evidence), sperm DNA studies
Fetal medicine	Rapid genome sequencing in critically ill neonates; fetal metabolomics in FGR
Newborn screening	Tandem MS of bloodspots (PKU, MCADD and other inborn errors)

237.4 Must-know summary

■ **EXAM TIP**

- Genome → epigenome → transcriptome → proteome → metabolome (closest to phenotype); microbiome adds environment.
- Key tools: **NGS**, microarray, **mass spectrometry**, NMR, bioinformatics.
- O&G: **NIPT (cfDNA)**, microarray/exome in anomalies, **sFlt-1/PlGF**, BRCA/HRD, Lynch/MMR, molecular endometrial cancer classification, vaginal microbiome and preterm birth.
- Challenges: VUS, validation, cost, ethics.

■ **MNEMONIC / MEMORY AID**

Omics order — 'Genes, Edits, Transcripts, Proteins, Metabolites, Microbes' (GET PM²).