BASIC & ADVANCED INVESTIGATIONS IN INFERTILITY

This presentation will cover the systematic approach to investigating infertility, from initial assessments to advanced diagnostic procedures. We will define infertility, detail basic and advanced investigations for both male and female partners, and conclude with key clinical considerations.

II. BASIC INFERTILITY INVESTIGATIONS

Female Partner: Initial Assessment & Diagnostics

History & Physical Examination

- Detailed menstrual, medical, surgical, and contraceptive history.
- Assessment of BMI, signs of endocrine dysfunction (e.g., PCOS, thyroid), and galactorrhea.

Ovulation Assessment

- Hormonal profile: Day 2/3 FSH, LH, E2 to assess ovarian function.
- Day 21 serum progesterone (level >3 ng/ml confirms ovulation).
- Ultrasound follicular monitoring and urinary LH kits for ovulation tracking.

Transvaginal Ultrasound (TVS)

- Evaluation of antral follicle count for ovarian reserve.
- Identification of uterine anomalies (e.g., fibroids, polyps) and endometrial thickness.

Thyroid & Prolactin

- TSH levels to rule out thyroid dysfunction (hypo/hyperthyroidism).
- Serum prolactin measurement to detect hyperprolactinemia, which can inhibit ovulation.

Hysterosalpingography (HSG)

- Performed in the follicular phase to assess fallopian tubal patency.
- Evaluates the uterine cavity for structural abnormalities.

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II. BASIC INFERTILITY INVESTIGATIONS

Male Partner: Essential Diagnostic Steps

Semen Analysis

The cornerstone of male infertility investigation. Conducted per WHO criteria, assessing volume, sperm count, motility, and morphology. Requires 2–3 days of abstinence and at least two samples for accuracy.

Hormonal Profile

Indicated for abnormal semen parameters or azoospermia. Includes FSH, LH, and Testosterone. Prolactin and TSH may be evaluated if clinical suspicion arises for endocrine imbalances affecting spermatogenesis.

Scrotal Ultrasound

Utilised to identify anatomical abnormalities such as varicoceles, assess testicular size, and detect potential obstructions within the reproductive tract. This imaging aids in localising the cause of male factor infertility.

III. ADVANCED INVESTIGATIONS

Female Partner: In-depth Diagnostics

Anti-Müllerian Hormone (AMH)

A reliable marker for ovarian reserve, reflecting the number of primordial follicles. Its levels are stable throughout the menstrual cycle, making it a convenient and accurate assessment.

Sonohysterography (SIS)

An enhanced ultrasound technique using saline infusion to distend the uterine cavity. Provides a clearer view for detecting intrauterine pathologies like polyps, septa, or adhesions that may impact implantation.

Diagnostic Hystero-Laparoscopy

A surgical procedure combining hysteroscopy (uterine cavity) and laparoscopy (pelvic organs). Allows for direct visualisation of tubal patency via chromopertubation, identification of endometriosis, adhesions, and subtle uterine anomalies that may not be visible on other scans.

Endometrial Biopsy

Seldom used, primarily for suspected chronic endometritis or assessing luteal phase defect, though its utility for the latter remains controversial. It involves a tissue sample from the uterine lining.

Thrombophilia & Autoimmune Screening

Considered in cases of recurrent pregnancy loss (RPL) or unexplained infertility. Screens for conditions that may lead to hypercoagulability or autoimmune responses affecting implantation and early pregnancy.

Genetic Testing (Karyotype)

Recommended for couples with recurrent pregnancy loss or premature ovarian insufficiency/failure in the female partner. Identifies chromosomal abnormalities that may contribute to reproductive issues.

III. ADVANCED INVESTIGATIONS

Male Partner: Specialised Tests

Sperm DNA Fragmentation Test

Measures the integrity of sperm DNA. High levels of DNA damage correlate with reduced fertilisation rates, poor embryo quality, and increased risk of recurrent pregnancy loss, even with normal routine semen parameters.

Genetic Testing

Includes karyotyping to identify chromosomal abnormalities (e.g., Klinefelter syndrome) and Y-chromosome microdeletion analysis, particularly relevant in men with azoospermia or severe oligozoospermia to detect genetic causes of impaired sperm production.

Testicular FNAC / Biopsy

Performed in men with azoospermia to distinguish between obstructive (sperm present in testes but blocked) and non-obstructive (impaired sperm production) causes. Guides further management, including sperm retrieval techniques.

Advanced Semen Tests

Includes assessment of Reactive Oxygen Species (ROS), which can indicate oxidative stress damaging sperm. The Hypo-osmotic Swelling Test assesses sperm membrane integrity and viability, providing insight beyond standard motility measures.

IV. Summary

Comprehensive Infertility Investigation

Hormonal Panel	FSH, LH, E2, AMH, TSH, Prolactin	FSH, LH, Testosterone, Prolactin
Imaging	TVS, HSG, SIS, Laparoscopy	Scrotal USG
Ovulation Testing	Serum Progesterone, USG, LH Kits	-
Genetic Tests	Karyotyping	Karyotyping, Y-Microdeletion
Other	Endometrial biopsy, Thrombophilia	DNA Fragmentation, ROS

V. Clinical Pearls

Key Considerations in Infertility Management

Simultaneous Evaluation

Initiate basic evaluation for both partners concurrently to expedite diagnosis and treatment. This integrated approach ensures no time is lost and provides a holistic view of the couple's fertility status.

Timely Advanced Testing

Avoid unnecessary delays in pursuing advanced investigations, especially in women over 35 or when the history strongly suggests significant pathology. Early intervention can preserve ovarian reserve and improve outcomes.

Holistic Counselling

Always counsel couples together, providing clear explanations of diagnoses, treatment options, and prognoses. Address the significant emotional and psychological impact of infertility, offering support and resources.

Thank you

We have explored the comprehensive spectrum of infertility investigations, from foundational assessments to highly specialised diagnostic procedures for both male and female partners.

Understanding these pathways is crucial for effective diagnosis and management, ultimately guiding couples towards appropriate and timely interventions.