Genetic Evolution Tournament

2025-10-29

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Preface

The Genetic Evolution Tournament (GET) is a Metaculus human judgment forecasting tournament established to generate forecasts and scenarios pertaining to the use of genetic and reproductive technologies on humans for treatment and enhancement.

Figure 1: From Wikipedia (2024-10-16): *Before the play begins, Kronos, the ruler of the pre-Olympian gods (the Titans), had been overthrown by an insurgency led by Zeus. In that revolt, Prometheus had sided with Zeus. As the new king, Zeus intended to destroy and replace humankind. Prometheus frustrated this plan, showing humans the use of fire, which Prometheus had stolen. Prometheus also taught humanity the arts. For these acts of defiance, Zeus intends to punish Prometheus by chaining him to a rock in the mountains of Scythia.*

1 About

This section comes directly from the repository's README…

> *The Genetic Evolution Tournament (GET) is a Metaculus human judgment forecasting tournament established to generate forecasts and scenarios pertaining to the use of human genetic and reproductive technologies for treatment and enhancement.*

Hosting

- GET is being hosted publicly on Metaculus.

Contact:

- Primary Administrator: (O957) + (@) + (proton) + (.) + (me)
- Secondary Administrator: (keystroke) + (@) + (duck) + (.) + (com)

On Feedback: All feedback is welcome; however, feedback that enhances

- (1) the accuracy of claims made in this tournament's questions (and other files)
- (2) the visibility and or utility of this tournament
- (3) other exact properties of the tournament (such as an additional question or an alteration in the structure)

is *particularly* desired.

1.1 Summary Introduction

- A Metaculus tournament.
- Focuses on human enhancement and treatment via genetic and reproduction technologies.
- Around 150 questions across 5 categories
  - Ethical, Legal, and Societal Implications
  - Technological Advancements and Clinical Applications
  - Regulatory and Policy Developments
  - Accessibility and Demographic Shifts
  - Healthcare and Insurance System Evolution
- At present, 15K USD in prizes
- Tentative end date of 2045

1.2 Extended Introduction

Disclaimer: *This description, which possibly contains inadequacies, is a minimally viable placeholder. Beyond the content detailing GET, there are many unsubstantiated claims. At some point, this tournament might be published as research, in which case the author will more strongly evidence claims regarding the landscape of current capabilities for human enhancement and treatment via genetic and reproductive means. GET questions often contain references to research literature, but these are far from comprehensive. The Extended Introduction section is liable to change.*

Overview: *This introduction starts with some motivating remarks on possible future(s) for human reproduction and human genetic constitution. Following this, the author briefly describes GET, which is currently under development and due to be released on Metaculus (as a public tournament).*

Scientific progress in genetics, genetic engineering, and assisted-reproduction continues at a rapid and accelerating pace, with current technological capabilities far exceeding those imagined by researchers and policy-makers in biotechnology at the start of the 21st century.

Frontier events in genetic engineering, such as the Jiankui He affair, have required all of humanity to consider scenarios and possibilities—ranging the gamut of expected value from fantastical to catastrophic—concerning the manipulation of human biology.

Although extreme-fidelity human gene modification (as in precisely exacting desired genetic outcomes) or, more broadly, modification of human biology at a caliber similar to what we might expect to be employed in *Brave New World* or *Gattaca*, is yet unrealized, developments in the aforementioned fields steer evermore towards this capacity.

Fortunately, considerable anticipatory work in ethics and governance have coincided with scientific and technological progress in genetic and reproductive research fronts. Ethical work on directed use of technology on human biology has involved some degree of implicit and or explicit scenario analysis, but this analysis (in the author's sampling of the literature) has infrequently employed forecasting.

Handling anticipatory blind spots and reducing uncertainty on futures involving humanity's possible genetic evolution via forecasting seems important for coordination between researchers, the public, and policy-makers in directing outcomes and safeguarding certain aspects of humanity's future.

Despite the expected outcome for GET being that most community traction will occur within the Effective Altruism, Less Wrong, and Metaculus communities, GET seeks to occupy the general niche of uncertainty reduction on futures involving varying levels of human-targeted DNA and assisted-reproduction technologies.

GET will ideally contribute, via the forecasts collected across its lifespan, to a broader scenario modelling effort occurring across discussions on the future(s) of human genetics and reproduction. These forecasts and their analysis might inform individual and policy decision-making on the technologies and procedures involved in human genetics and reproduction.

With respect to the forecasting platform for GET, given both Metaculus's status as a reputable human judgement forecast (HJF) aggregator (this is the author's impression) and the author's familiarity with the site (the author is biased in this way), Metaculus was chosen as the site to host GET. The particular language and norms adopted in public tournaments on Metaculus typically incentivize forecasters to make clear and well-reasoned comments and to forecast as honestly and as accurately as possible. For example Metaculus tournaments, see this page.

GET currently consists of 5 question categories:

- (1) Ethical, Legal, and Societal Implications
- (2) Technological Advancements and Clinical Applications
- (3) Regulatory and Policy Developments
- (4) Accessibility and Demographic Shifts
- (5) Healthcare and Insurance System Evolution

Each category was intended to have between 15 and 25 questions; the intention was to between 75 to 125 questions in total.

To incentivize forecaster participation, there are prizes totaling 15K USD, though this may increase in the future. Prizes are partitioned into a forecasting accuracy component, with questions scored via Metaculus's default scoring procedures (peer scores), and a commenting component.

Originally, within the commenting component, 2.5k USD was allocated to comments on questions that are part of the forecasting accuracy component (shorter-term questions) and 7.5k USD was allocated to comments on longer-term questions. However, after some deliberation, a decision was made to break the commenting section of the tournament into 3 stages, each with 2k USD. The tournament is expect to last roughly 20 years. There is the possibility for additional funding altering the structure of the tournament.

2 Goals

GET has, broadly, the following goals:

- Generate questions on possible **civilizational impacts** of current and near term human genetic and reproductive technologies.
- Generate probabilistic **human-judgment forecasts** on the questions in the previous point.
- Synthesize the probabilistic forecasts in the previous point to make **quantitative statements on possible futures** (scenarios) for humanity involving the use of human genetic and reproductive technologies, with an emphasis on human enhancement.
- Communicate the questions, forecasts, and statements in previous points via Internet posts and or **publication(s)**.

Administrators of GET seek to achieve these goals by:

...Section pending, as this depends on the existence of forecasts and the feedback of experts and forecasters

3 Hosting

GET is publicly hosted on Metaculus:

- Metaculus GET Page: https://www.metaculus.com/tournament/genetic/?

GET also has a GitHub page and a website:

- GET Website: https://genetictournament.com/
- GET GitHub Page: https://github.com/O957/Genetic-Evolution-Tournament

4 Administration

GET is administered by:

- H559 (primary administrator)
- James Evans (secondary administrator)

Administration includes:

- Answering participants' questions about GET.
- Resolving GET forecasting questions on Metaculus.
- Allocating prize funds to GET prize holders.
- Continuing the tournament if Metaculus ceases to exist.
- Writing new questions and updating existing questions.
- Announcement of GET launch and new stages.
- Analysis and communication of forecasts produced.

To contact administrators directly:

- Primary Administrator: (O957) + (@) + (proton) + (.) + (me)
- Secondary Administrator: (keystroke) + (@) + (duck) + (.) + (com)

5 Contact

To contact administrators directly:

- Primary Administrator: (O957) + (@) + (proton) + (.) + (me)
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6 Structure

Overview: *The author first explains how GET is set up, temporally speaking. Next, the author explains the size of GET's prize pool. The author closes with considerations for how these aforementioned topics may change.*

Disclaimer: *If this disclaimer is present, then the below sections are still being developed. Suggestions here would very much be appreciated. Please either make an issue here, contact the author, or fill out this form.*

GET is set up in stages:

- Tournament releases early 2025, estimate January 15th.
- *Short-Term Insights* section begins 2025-01-15, lasts until 2029-12-31.
- *Medium-Term Insights* section begins 2030-01-01, lasts until 2034-12-31.
- *Long-Term Insights* section begins 2035-01-01, lasts until 2045-01-01.
- Questions that do not resolve before 2045-01-01 are removed tournament scoring, but not from Metaculus.

Notes on the stages:

- GET is expected to initially have around 150 questions across 5 categories.
- Future questions will likely be added, especially as need arises[1].
- Different questions have different hidden[2] periods, based on their close dates.
- (critical) Questions belong to the stage in which they resolve or close.

Prize structure:

- GET is currently (2024-11-21) funded at PRIZE = 15K USD.
- The prize pool might increase at some point in the future.
- All stages have a commenting prize pool of 2/15 x PRIZE.
- All stages have 10 commenting prizes.
- Awardable comment periods:

[1] For example, new technologies or questions conditional on other questions resolving.

[2] A hidden period on Metaculus is when the Community Forecast is not visible to forecasters. This allows for non-autocorrelated forecasting for at least some period of the question.

  - *Short-Term Insights*: [2025-01-01, 2027-12-31]
  - *Medium-Term Insights*: [2025-01-01, 2032-12-31]
  - *Long-Term Insights*: [2025-01-01, 2032-12-31]

- Awards for comments in each stage are awarded at the end of the stage.
- Forecasting accuracy prize percentages by stage:

  - *Short-Term Insights*: 3.25/15 x PRIZE
  - *Medium-Term Insights*: 2.5/15 x PRIZE
  - *Long-Term Insights*: 1.75/15 x PRIZE

- Forecasting accuracy awards for each stage are also awarded at the end of the stage; these awards cover questions that resolved early i.e. before the stage that contained their close date.
- Conditional on certain questions resolving ambiguously within a stage, some such questions might be created again for a later stage, should the community generally express interest in this.

Prize restrictions:

- No one participant may receive greater than 1K USD in commenting prizes.
- Commenting prizes will only be distributed within a stage conditional on there being at least 5 comments.
- Commenting prize money not allocated with a stage will be
- Administrators may not receive prizes on comments but may receive forecasting accuracy prizes.

Comment award criteria[3]:

- The comment pertains to correct time horizons
- The comment is well-reasoned and clearly written
- The comment provides valuable information and reasoning
- The comment identifies mistakes in the community's reasoning or clearly explains why the author's forecast differs from the community's
- The comment describes connections between forecasts on different question groups, or inconsistencies between forecasts on different question groups

[3]At present, the author is deferred to the criteria used in the OWID tournament, which is, to some degree, qualitative.

7 Scoring

For GET commenting prizes:

- Evaluated by tournament administrators.
- Final commenting prize selections *may* be reviewed additionally by Metaculus administrators.
  - Should this occur, public notice will be provided.

For GET forecast scoring prizes:

- Metaculus scoring algorithms will be used.
- For information on Metaculus scoring, click here.

8 External Considerations

Concerning catastrophe:

> Should humanity go extinct* or experience a catastrophe* that results in the collapse of complex society* for over a continuous 5 year interval, all questions in the *Genetic Evolution Tournament* resolve as **Ambiguous**.

Concerning the non-existence of Metaculus:

- GET administrators will self-host the 2024-2025 open-source version of Metaculus.

9 Feedback

To provide feedback, use one of the four avenues:

- GitHub Discussion Comment
- GitHub Issue
- Google Feedback Form
- Metaculus Discussion Comment

Part I

Q: Ethical, Legal, & Societal Implications

10 Introduction

11 Notes

- Government tax incentives for those using [gene therapy, polygenic screening, genetic modification]?
- Number of instances genetic disorders decrease?
  - which disorder?
  - when?
  - which nations?
  - due to? [PGS, genetic engineering]
-

Part II

Q: Technological Advancements & Clinical Applications

Part III

Q: Regulatory & Policy Developments

Part IV

Q: Accessibility & Demographic Shifts

12 Notes

- That which is needed for polygenic screening
  - Open source code available?
- That which is needed for genetic modification
  - Requires clinic?
- Cost of [gene therapy, genetic modification, polygenic screening]
  - cost of IVF?
  - cost of egg collection?
  - cost of sperm donations?
  - cost of screening?

By virtue of effective screening and or genetic modifications how will data collection change?

Unions against IVF, polygenic screening.

Backlash, protests.

How much suppressions.

Lobbying, non-profits

Number of open-source repositories on GitHub focusing on PGS.

Documentaries, number of documentaries. Movies database. IMDB.

Part V

Q: Healthcare & Insurance System Evolution

13 Notes

- Insurance coverage for?
  - Genetic modification
  - Gene therapy
  - Polygenic screening
- If insurance coverage, for [polygenic screening, gene therapy, genetic modification]
  - how selective?
  - how costly?
  - how different by country?
- Cost of insurance increase
  - if enhance modification?
  - if "poor choice" modification, e.g. aggression
- Cost of insurance decrease
  - if chose certain modification, e.g. moral enhancement
- Healthcare costs associated predominantly
  - if someone was screened, because they were screened
  - if someone was not screened, because they were screened

Part VI

Announcements

14 LW & EAF

15 Elsewhere

Part VII

Resources

16 Scholarly Articles

17 Books

The author, in working on this tournament, has found the following books useful. At some point, links might be added connecting the author's notes to these books.

17.1 Choosing Children: Genes, Disability, And Design

Link: https://academic.oup.com/book/11973

Citation:

> Glover, Jonathan. Choosing children: Genes, disability, and design. Oxford University Press, 2006.

Bibtex:

```
@book{glover2006choosing,
  title={Choosing children: Genes, disability, and design},
  author={Glover, Jonathan},
  year={2006},
  publisher={Oxford University Press}
}
```

17.2 Clinical ethics at the crossroads of genetic and reproductive technologies

Link: https://www.sciencedirect.com/book/9780443190452/clinical-ethics-at-the-crossroads-of-genetic-and-reproductive-technologies

Citation:

> Hostiuc, Sorin, ed. Clinical ethics at the crossroads of genetic and reproductive technologies. Elsevier, 2023.

Bibtex:

```
@book{hostiuc2023clinical,
  title={Clinical ethics at the crossroads of genetic and reproductive technologies},
  author={Hostiuc, Sorin},
  year={2023},
  publisher={Elsevier}
}
```

17.3 Creating Future People: The Ethics Of Genetic Enhancement

Link: https://library.oapen.org/handle/20.500.12657/58965

Citation:

> Anomaly, Jonathan. Creating future people: The ethics of genetic enhancement. Taylor & Francis, 2020.

Bibtex:

```
@book{anomaly2020creating,
  title={Creating future people: The ethics of genetic enhancement},
  author={Anomaly, Jonathan},
  year={2020},
  publisher={Taylor \& Francis}
}
```

17.4 Enhancing Human Capacities

Link: https://onlinelibrary.wiley.com/doi/book/10.1002/9781444393552

Citation:

> Savulescu, Julian, Ruud ter Meulen, and Guy Kahane. "Enhancing Human Capacities." (2011).

Bibtex:

```
@article{savulescu2011enhancing,
  title={Enhancing Human Capacities},
  author={Savulescu, Julian and ter Meulen, Ruud and Kahane, Guy},
  year={2011},
  publisher={Wiley Online Library}
}
```

17.5 The End Of Sex And The Future Of Human Reproduction

Link: https://www.hup.harvard.edu/books/9780674984011

Citation:

> Greely, Henry T. The end of sex and the future of human reproduction. Harvard University Press, 2016.

Bibtex:

```
@book{greely2016end,
  title={The end of sex and the future of human reproduction},
  author={Greely, Henry T},
  year={2016},
  publisher={Harvard University Press}
}
```

18 LW & EAF Posts

19 Wikipedia

20 Glossary

This document contains several collected glossaries across.

Online Sites:

- Glossary of Genetic Engineering Terms by Dr. Mae-Wan Ho.
- Assisted Reproductive Technology (ART) Glossary Of Terms by The CDC.
- Assisted Reproductive Technology - Glossary UNSW Embryology
- Society For Assisted Reproductive Technology
- The Internal Glossary on Infertility and Fertility Care by International Committee Monitoring Assisted Reproductive Technologies
- Glossary of Terms by CDC Genomics
- NCI Dictionary of Genetics Terms by National Cancer Institute

Books:

- Creating Future People
- The End Of Sex And The Future Of Human Reproduction

aCGH Array

- Comprehensive genomic hybridization, sometimes abbreviated ACGH or array CGH, is a laboratory technique that can detect aneuploidies (samples with the wrong number of chromosomes) as well as much smaller deletions, duplications, or movements of DNA within or between chromosomes.

acrochordon

- A small, benign skin growth that may have a stalk (peduncle). Acrochordons most commonly appear on the neck, axillary, groin, and inframammary regions. Also called skin tag.

Acrosome

- A membrane-bound structure covering the anterior of the sperm head that contains enzymes necessary to penetrate the zona pellucida of the oocyte.

Adaptive mutation, or Directed mutation

- The phenomenon whereby bacteria and yeast cells in stationary (non-growing) phase, have some way of producing (or selectively retaining) only the most appropriate mutations that enable them to make use of new substrates for growth.

Adenomyosis

- A form of endometriosis marked by the presence of endometrium-like epithelium and stroma outside the endometrium in the myometrium.

Adhesions

- Bands of fibrous scar tissue that may bind the abdominal and pelvic organs, including the intestines and peritoneum, to each other. They can be dense and thick or filmy and thin.

affected individual

- In genetics, describes an individual who has a particular phenotypic trait or disease. The individual expresses the trait or has signs and symptoms of the disease.

Age Specific Fertility Rate (ASFR)

- The number of live births per woman in a particular age group in a specific calendar year expressed per 1000 women in that age group.

Agglutination

- Clumping of spermatozoa in the ejaculate.

Allele

- A particular variant of a gene.
- An allele is the version of the gene that is present. Each person has two alleles for each gene, one from each parent. If the alleles of a gene are the same, the person is homozygous for the gene. If the alleles are different, the person is heterozygous for the gene.
- One variant of a gene. A gene may have many different alleles, each with a slightly different DNA sequence. Some may be harmful, some helpful; some may make neutral changes, and many will make no discernible change. The most common allele is sometimes called the 'wild type' allele.

allele

- One of two or more versions of a genetic sequence at a particular region on a chromosome. An individual inherits two alleles for each gene, one from each parent.

allelic heterogeneity

- The presence of different variants at a single gene locus that cause the same or similar phenotypic expressions of a disease or condition.

Allergen

- A substance that causes the body to react hypersensitively to it.

Alpha-fetoprotein

- A protein produced by a human fetus. Some of it crosses the placenta and can be found in the pregnant woman's blood serum, where its levels help predict Down syndrome or neural tube defects.

always cooperate

- Cooperate with other players no matter what they do.

always defect

- Defect against other players no matter what they do.

American Society for Reproductive Medicine (ASRM)

- A professional medical organization of more than 8,000 health care professionals dedicated to reproductive medicine.

Amniocentesis

- A procedure in which a small amount of amniotic fluid is removed through a needle from the fetal sac at about 16 weeks into a pregnancy. The fluid is studied for chromosomal or other abnormalities which may affect fetal development.

Andrology

- The medical practice dealing with the health of the male reproductive system.

Aneuploid

- A cell that has the wrong number of chromosomes. Human cells should normally have 46 chromosomes (92 when preparing to divide). Trisomies, which are three copies of what should be a paired chromosome, are the most common aneuploidies in living humans.

Aneuploidy

- An abnormal number of chromosomes in a cell. The majority of embryos with aneuploidies are not compatible with life.

aneuploidy

- The occurrence of one or more extra or missing chromosomes in a cell or organism. Aneuploidy refers to any chromosome number that is not an exact multiple of the haploid number of chromosomes (which is 23 in humans).

Anti-müllerian Hormone (AMH)

- A hormone which is often measured in a woman to help determine her egg supply, or 'ovarian reserve'. It is secreted by small, growing follicles.

Anti-sperm antibodies

- Antibodies that recognize and bind to antigens on the surface of the spermatozoon.

Antibiotic resistance marker genes

- Genes coding for antibiotic resistance used in genetic modification. They allow the cells that have taken up the foreign GM construct to be selected with antibiotics, and frequently remain in the genetically modified organism and transgenic line created.

antioncogene

- A type of gene that regulates cell growth. When an antioncogene is mutated, uncontrolled cell growth may occur. This may contribute to the development of cancer. Also called tumor suppressor gene.

Antral follicle count

- The number of follicles noted by ultrasound at the beginning of the menstrual cycle, usually day 2 or 3.

ART

- Procedures that use donor or nondonor eggs and sperm to create embryos in vitro. Examples of ART include in vitro fertilization (IVF), gamete intrafallopian transfer (GIFT), and zygote intrafallopian transfer (ZIFT). These procedures may be used prior to preimplantation genetic testing (PGT). Also called assisted reproductive technology.
- See Assisted reproductive technology.

ART (assisted reproductive technology)

- All treatments or procedures that include the handling of human eggs or embryos to help achieve a pregnancy. The most common type of ART is in vitro fertilization (IVF). Other types of ART, such as gamete intrafallopian transfer or zygote intrafallopian transfer, are rarely performed. ART also includes egg and embryo cryopreservation (freezing) and egg and embryo donation. An ART procedure is sometimes referred to as a cycle of treatment or an ART cycle.

Ashkenazi Jews

- One of two major ancestral groups of Jewish people whose ancestors lived in France and Central and Eastern Europe, including Germany, Poland, and Russia. The other group is called Sephardic Jews and includes those whose ancestors lived in Spain, Portugal, North Africa, and the Middle East. Most Jewish people living in the United States are of Ashkenazi descent.

Aspermia

- Lack of external ejaculation.

ASRM

- The American Society for Reproductive Medicine, a nonprofit organization dedicated to reproductive medicine. It has an affiliate for reproductive medicine clinics called SART (Society for Assisted Reproductive Technologies).

Assisted hatching

- An ART procedure in which the zona pellucida of an embryo is either thinned or perforated by chemical, mechanical or laser methods.
- An in vitro procedure in which the zona pellucida of an embryo is either thinned or perforated by chemical, mechanical, or laser methods to assist separation of the blastocyst.

Assisted hatching (AH)

- A procedure in which the zona pellucida (outer covering) of the embryo is partially opened, usually by application of an acid or laser, to facilitate embryo implantation and pregnancy.

Assisted reproductive technologies (ART)

- All treatments which include the handling of eggs and/or embryos. Some examples of ART are in vitro fertilization (IVF), gamete intrafallopian transfer (GIFT), pronuclear stage tubal transfer (PROST), tubal embryo transfer (TET), and zygote intrafallopian transfer (ZIFT).

assisted reproductive technology

- Procedures that use donor or nondonor eggs and sperm to create embryos in vitro. Examples include in vitro fertilization (IVF), gamete intrafallopian transfer (GIFT), and zygote intrafallopian transfer (ZIFT). These procedures may be used prior to preimplantation genetic testing (PGT). Also called ART.

Assisted reproductive technology

- (ART) All treatments or procedures that include the in vitro handling of both human oocytes and sperm or of embryos for the purpose of establishing a pregnancy. This includes, but is not limited to, in vitro fertilization and embryo transfer, gamete intrafallopian transfer, zygote intrafallopian transfer, tubal embryo transfer, gamete and embryo cryopreservation, oocyte and embryo donation, and gestational surrogacy. ART does not include assisted insemination (artificial insemination) using sperm from either a woman's partner or a sperm donor. (More? Assisted Reproductive Technology)

Assisted reproductive technology (ART)

- All interventions that include the in vitro handling of both human oocytes and sperm or of embryos for the purpose of reproduction. This includes, but is not limited to, IVF and embryo transfer (ET), intracytoplasmic sperm injection (ICSI), embryo biopsy, preimplantation genetic testing (PGT), assisted hatching, gamete intrafallopian transfer (GIFT), zygote intrafallopian transfer, gamete and embryo cryopreservation, semen, oocyte and embryo donation, and gestational carrier cycles. Thus, ART does not, and ART-only registries do not, include assisted insemination using sperm from either a woman's partner or a sperm donor. (See broader term, medically assisted reproduction, MAR.)

Asthenoteratozoospermia

- Reduced percentages of motile and morphologically normal sperm in the ejaculate below the lower reference limit. When reporting results, the reference criteria should be specified.

Asthenozoospermia

- Reduced percentage of motile sperm in the ejaculate below the lower reference limit. When reporting results, the reference criteria should be specified.

attributable risk

- Proportion of a disease in exposed individuals that can be attributed to an exposure. In genetic studies, the 'exposure' is the frequency of a specific genetic variant.

Autologous cycle

- Treatment cycles that use the women.s own oocytes or embryos.

autosomal

- Having to do with any of the 22 numbered pairs of chromosomes found in most human cells. Autosomal chromosomes are numbered 1–22. The sex chromosomes (X and Y) are not autosomal.

autosomal dominant inheritance

- A mode of inheritance in which a genetic condition occurs when a variant is present in only one allele (copy) of a given gene.

autosomal recessive inheritance

- A mode of inheritance in which a genetic condition occurs when a variant is present on both alleles (copies) of a given gene.

Autosome

- A chromosome other than the sex-chromosome, or sex-determining chromosome.
- One of the chromosomes that is not a sex chromosome. In humans there are 22 pairs of autosomes, named chromosomes 1 through 22.

Azoospermia

- The absence of spermatozoa in the ejaculate.

Bacteriophage

- Any virus that infect bacteria, also known as phage.

band

- A pattern of light and dark stripes on chromosomes produced by staining during cell division; used to assign chromosome numbers and evaluate structure.

base pair

- Two nitrogen-containing bases (nucleotides) that pair together to form the structure of DNA. The four bases in DNA are adenine (A), cytosine (C), guanine (G), and thymine (T). The term can also refer to the number of base pairs in a DNA sequence.

behavior genetics

- The study of how genes interact with the environment to influence behavior.

biallelic

- Pertaining to both alleles of a single gene (one paternal and one maternal). For example, biallelic mutation carriers have mutations in both copies of a gene.

Binucleation

- The presence of two nuclei in a blastomere (cell).

Biochemical pregnancy

- (preclinical spontaneous abortion/ miscarriage) A pregnancy diagnosed only by the detection of hCG in serum or urine and that does not develop into a clinical pregnancy.
- When a woman's pregnancy test is initially positive but becomes negative before a gestational sac is visible on ultrasound.

Biochemical Pregnancy

- A pregnancy diagnosed only by the detection of beta hCG in serum or urine.

Biofilm

- A layer of extracellular matrix containing quiescent, non-proliferating micro-organisms.

Birth (single)

- The complete expulsion or extraction from a woman of a fetus after 22 completed weeks of gestational age, irrespective of whether it is a live birth or stillbirth, or, if gestational age is unknown, a birth weight more than 500 grams. A single birth refers to an individual newborn; and a delivery of multiple births, such as a twin delivery, would be registered as two births.

Blastocoele

- Fluid-filled central region of the blastocyst.

Blastocyst

- A human embryo from the fifth day after fertilization until implantation. It takes the form of a hollow sphere containing the inner cell mass.
- An embryo that has formed a fluid-filled cavity and the cells have begun to form the early placenta and embryo, usually 5 days after ovulation or egg retrieval.
- An embryo, 5 or 6 days after fertilization, with an inner cell mass, outer layer of trophectoderm, and a fluid-filled blastocele cavity.
- The stage of preimplantation embryo development that occurs around day 5–6 after insemination or ICSI. The blastocyst contains a fluid filled central cavity (blastocoele), an outer layer of cells (trophectoderm) and an inner group of cells (inner cell mass).

Blastomere

- A cell in a cleavage stage embryo.

Blastomere Symmetry

- The extent to which all blastomeres are even in size and shape.

Bleeding after oocyte aspiration

- Significant bleeding, internal or external, after oocyte aspiration retrieval requiring hospitalization for blood transfusion, surgical intervention, clinical observation or other medical procedure.

Canceled ART cycle

- An ART cycle in which ovarian stimulation or monitoring has been initiated with the intention to treat, but which did not proceed to follicular aspiration or in the case of a thawed or warmed embryo did not proceed to embryo transfer.

Canceled cycle

- An ART cycle in which ovarian stimulation or monitoring has been carried out with the intention to treat, but which did not proceed to follicular aspiration or, in the case of a thawed embryo, to embryo transfer.

carrier

- In classical genetics, an individual who carries one deleterious allele for an autosomal recessive disorder; clinically, may refer to someone carrying a deleterious allele that predisposes to disease.

carrier frequency

- The proportion of individuals in a population who have a single copy of a specific recessive genetic variant. Also called carrier rate.

carrier rate

- The proportion of individuals in a population who have a single copy of a specific recessive genetic variant. Also called carrier frequency.

cascade genetic testing

- The process of extending genetic testing to at-risk family members for a pathogenic variant identified in a relative; also called cascade testing.

cascade screening

- A systematic process for identifying individuals within a family at risk for a hereditary condition by testing for a known family variant; sometimes called cascade testing.

cascade testing

- The process of extending genetic testing to at-risk family members for a pathogenic variant identified in a relative; also called cascade genetic testing.

Cell line

- A supposedly genetically uniform population of cells derived from one individual cell. The genetic identity of all the cells is a fiction, as the genetic material is subject to many 'fluid genome' processes that constantly make cells genetically different from one another, and especially in culture. Both plant and animal cells are subject to large variations known collectively as somaclonal variations.

Centers for Disease Control and Prevention (CDC)

- Federal agency for protecting the health and safety of people at home and abroad, providing credible information to enhance health decisions, and promoting health through strong partnerships.

Centromere

- The constricted part of a chromosome between its p (short) and q (long) arms, essential for proper allocation of chromosomes to daughter cells during cell division.

Cervical canal

- The passageway leading from the vagina into the uterus.

Cervical mucus

- The substance in the cervix through which sperm must swim to enter the uterus.

Cervix

- The narrow, lower end of the uterus.

Childlessness

- A condition in which a person, voluntarily or involuntarily, is not a legal or societally-recognized parent to a child, or has had all children die.

Chimerism

- Presence in a single individual of two or more cell lines, each derived from different individuals.

CHIP

- Clonal hematopoiesis of indeterminate potential: the presence of somatic mutations in hematopoietic stem cells in an individual without detectable hematologic cancer, with a variant allele frequency of 2%, associated with increased risk of cardiovascular and hematologic diseases.

chromosomal instability

- A genomic imbalance occurring when a cell has an abnormal number of chromosomes, caused by unexpected chromosomal crossover or extra-chromosomal DNA pieces.

chromosome

- A structure found inside the nucleus, composed of DNA and proteins, organized into genes. Each human cell normally contains 23 pairs of chromosomes.

Chromosome

- A long DNA molecule wrapped around a protein backbone that carries the genetic information, which is duplicated and allocated to daughter cells during cell division.
- A structural unit of genetic material consisting of a long molecule of DNA complexed with special proteins in eukaryotes, but not in prokaryotes
- DNA is packaged into small units called chromosomes. A chromosome contains a single, long piece of DNA with many different genes. Every human cell contains 23 pairs of chromosomes. There are 22 pairs of numbered chromosomes, called autosomes, and one pair of sex chromosomes, which can be XX or XY. Each pair contains two chromosomes, one from each parent, which means that children get half of their chromosomes from their mother and half from their father.

Cleavage Stage Embryos

- Embryos beginning with the 2-cell stage and up to, but not including, the morula stage.

CLIA

- The Clinical Laboratories Improvements Amendments of 1988, which regulate clinical laboratories in the US through agencies like the FDA, CDC, and CMS.

Clinical fertility

- The capacity to establish a clinical pregnancy.

Clinical pregnancy

- A pregnancy confirmed by an increasing level of hCG and the presence of a gestational sac detected by ultrasound.
- A pregnancy diagnosed by ultrasonographic visualization of one or more gestational sacs or definitive clinical signs of pregnancy. In addition to intra-uterine pregnancy, it includes a clinically documented ectopic pregnancy.
- A pregnancy diagnosed by ultrasonographic visualization of one or more gestational sacs or definitive clinical signs of pregnancy. It includes ectopic pregnancy. Note: Multiple gestational sacs are counted as one clinical pregnancy.

- A pregnancy documented by ultrasound that shows a gestational sac (fluid-filled structure that develops early in pregnancy surrounding the embryo) in the uterus. For ART data-reporting purposes, pregnancy is defined as a clinical pregnancy rather than a chemical pregnancy (positive pregnancy test).

Clinical pregnancy rate

- The number of clinical pregnancies expressed per 100 initiated cycles, aspiration cycles or embryo transfer cycles. When clinical pregnancy rates are recorded, the denominator (initiated, aspirated or embryo transfer cycles) must be specified.
- The number of clinical pregnancies expressed per 100 initiated cycles, aspiration cycles, or embryo transfer cycles. Note: When clinical pregnancy rates are given, the denominator (initiated, aspirated, or embryo transfer cycles) must be specified.

Clinical pregnancy with fetal heart beat

- A pregnancy diagnosed by ultrasonographic or clinical documentation of at least one fetus with a discernible heartbeat.
- Pregnancy diagnosed by ultrasonographic or clinical documentation of at least one fetus with heart beat. It includes ectopic pregnancy.

clinical utility

- The likelihood that a test will prompt an intervention that results in improved health outcomes.

clinical validity

- The predictive value of a test for a specific clinical outcome, determined by its sensitivity and specificity.

Clomiphene citrate

- An oral antiestrogen medication used to induce ovulation.

Clomiphene citrate challenge test (CCCT)

- A test of ovarian reserve in which serum FSH is checked on days 3 and 10 of the menstrual cycle and clomiphene citrate is taken on days 5 through 9.

clonal hematopoiesis of indeterminate potential

- The presence of somatic mutations in hematopoietic stem cells in an individual without detectable hematologic cancer, with mutations at a variant allele frequency of 2%; also called CHIP.

Clone

- An identical copy of an individual organism, a cell, or a gene, or the totality of all the identical copies made from an individual organism, a cell, or a gene. In genetics, the clone implies identical in genetic make-up to the original.

Clone/cloning

- A clone is an exact copy of something, and cloning is the process of making such copies. In biology, it generally refers to nonsexual reproduction producing offspring with the same DNA as their progenitors. Identical twins are natural clones, and somatic cell nuclear transfer (SCNT) is a cloning technique in mammals.

CNV

- A copy number variant; a variation in the number of copies of a specific DNA sequence, including insertions, deletions, or duplications.

CNVs

- Copy number variations are regions in DNA where some individuals have more or fewer copies of a particular gene sequence than normal, due to duplications or deletions.

codon

- A sequence of three consecutive nucleotides in DNA or RNA that codes for a specific amino acid, including start and stop signals.

Codon

- Three 'letters' or bases of DNA or RNA that code for an amino acid, or signal start or stop during protein synthesis.

Cohort total fertility rate (CTFR)

- The observed average number of live born children per woman applied to a birth cohort of women as they age through time. This is obtained from data on women after completing their reproductive years.

collective action problem

- A situation in which each person acting rationally, according to their own goals, produces a worse outcome than they would if all acted in a different way.

Compaction

- The process during which tight junctions form between juxtaposed blastomeres resulting in a solid mass of cells with indistinguishable cell membranes.

Complex aneuploidies

- Two or more aneuploidies involving different chromosomes in the embryo. When autosomes are involved, this condition is not compatible with human life.

compound heterozygosity

- The presence of two different mutated alleles at a particular gene locus.

conditionally cooperate

- Cooperate if and only if your opponent cooperates.

congenital

- Present at birth; a condition that may be caused by genetic and/or non-genetic factors.

Congenital anomalies

- All structural, functional, and genetic anomalies diagnosed in aborted fetuses, at birth, or in the neonatal period.
- Structural or functional disorders that occur during intra-uterine life and can be identified prenatally, at birth or later in life. Congenital anomalies can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. The time of identification should be reported.

Congenital anomaly birth rate

- The number of births exhibiting signs of congenital anomalies per 10,000 births. The time of identification should have been reported.

Congenital bilateral absence of the vasa deferentia (CBAVD)

- The absence, at birth, of both duct systems (vas deferentia) that connect the testes to the urethra and may be associated with cystic fibrosis transmembrane conductance regulator (CTFR) gene mutation. Although the testes usually develop and function normally, men present with azoospermia.

Conjugation

- The mating process in bacteria which require cell to cell contact being established, and in which genes are transferred between cells.

consanguinity

- Genetic relatedness between individuals who share a common ancestor.

consent process

- An information exchange between a clinician and an individual (or proxy) to facilitate informed decision making, including discussion of risks, benefits, and privacy regarding genetic testing; also called informed consent.

constitutional DNA

- Tissue-derived DNA from reproductive cells (egg or sperm) that is incorporated into every cell of an offspring; also called germline DNA.

consultand

- An individual who presents for genetic counseling.

Controlled ovarian stimulation (COS) for ART

- Pharmacologic treatment in which women are stimulated to induce the development of multiple ovarian follicles to obtain multiple oocytes at follicular aspiration.

Controlled ovarian stimulation (COS) for non-ART cycles

- Pharmacologic treatment for women in which the ovaries are stimulated to ovulate more than one oocyte.

Conventional in vitro insemination

- The co-incubation of oocytes with sperm in vitro with the goal of resulting in extracorporeal fertilization.

copy number variant

- A variation in the number of copies of a particular DNA sequence in the genome; also called CNV.

Copy Number Variation (CNV)

- A copy number variation (CNV) is when the number of copies of a gene or other section of DNA is different between people.

Corona radiata cells

- The innermost cells of the cumulus oophorus.

COS

- Acronym for Controlled Ovarian Stimulation. (More? Controlled Ovarian Stimulation)

cosegregation

- The joint transmission of two or more genes on the same chromosome due to close physical proximity (linkage).

CRISPR

- Short for Clustered Regularly Interspaced Short Palindromic Repeats. CRISPR allows bacteria to sequence and disable viruses that attack them. Because of its accuracy, scientists use a version of this system (usually CRISPR Cas9) to make precise cuts to DNA in order to alter genes, including the genes of human embryos.

Cross border reproductive care

- The provision of reproductive health services in a different jurisdiction or outside of a recognized national border within which the person or persons legally reside.

Cryopreservation

- Freezing at a very low temperature, such as in liquid nitrogen (196°C) to keep embryos, eggs, or sperm viable.
- The freezing or vitrification and storage of gametes, zygote, embryos, or gonadal tissue.
- The practice of freezing eggs or embryos from a patient's ART cycle for potential future use.
- The process of slow freezing or vitrification to preserve biological material (e.g. gametes, zygotes, cleavage-stage embryos, blastocysts or gonadal tissue) at extreme low temperature.

Cryopreserved

- Frozen.

Cryptorchidism

- Testis not in scrotal position within the neonatal period and, up to but not limited to, 1 year post birth. If the testis has not descended into the scrotum, this condition can cause primary testicular failure and increased risk of testicular cancer development.

Cumulative delivery rate per aspiration/initiated cycle with at least one live birth

- The number of deliveries with at least one live birth resulting from one initiated or aspirated ART cycle, including all cycles in which fresh and/or frozen embryos are transferred, until one delivery with a live birth occurs or until all embryos are used, whichever occurs first. The delivery of a singleton, twin, or other multiples is registered as one delivery. In the absence of complete data, the cumulative delivery rate is often estimated.

Cumulative delivery rate with at least one live born baby

- The estimated number of deliveries with at least one live born baby resulting from one initiated or aspirated ART cycle including the cycle when fresh embryos are transferred and subsequent frozen/ thawed ART cycles. This rate is used when less than the total number of embryos fresh and/or frozen/thawed have been used from one ART cycle. Note: The delivery of a singleton, twin, or other multiple pregnancy is registered as one delivery.

Cumulus oophorus

- The multi-layered mass of granulosa cells surrounding the oocyte.

cutaneous leiomyoma

- A benign tumor arising from smooth muscle tissue in a hair follicle, forming a papule that can be painful with cold or tactile stimuli.

cutaneous lichen amyloidosis

- Itchy, brownish-red papules typically appearing on the shins, thighs, feet, or neck.

cytogenetics

- The study of the structure, function, and abnormalities of human chromosomes.

Cytoplasmic maturation

- The process during which the oocyte acquires the capacity to support nuclear maturation, fertilization, pronuclei formation, syngamy and subsequent early cleavage divisions until activation of the embryonic genome.

Cytoplasmic transfer

- A procedure that can be performed at different stages of an oocyte's development to add to or replace various amounts of cytoplasm from a donor egg.

de novo mutation

- A genetic alteration present for the first time in a family member, resulting from a variant in a germ cell of a parent or in the fertilized egg itself; also called de novo variant, new mutation, or new variant.

de novo variant

- A genetic alteration present for the first time in a family member, resulting from a variant in a germ cell of a parent or in the fertilized egg itself; also called de novo mutation, new mutation, or new variant.

Decreased spermatogenesis

- A histological finding in which spermatogenesis is present with few cells in the seminiferous tubules, resulting in a decreased number or absence of sperm in the ejaculate.

Delayed ejaculation

- A condition in which it takes a man an extended period of time to reach orgasm and ejaculation.

Delayed embryo transfer

- A procedure in which embryo transfer is not performed within the time frame of the oocyte aspiration cycle but at a later time.

deleterious mutation

- A genetic alteration that increases an individual's susceptibility to disease; also called disease-causing mutation, pathogenic variant, predisposing mutation, or susceptibility gene mutation.

deletion

- A genetic change involving the loss of a segment of DNA, which can range from a single base to larger segments.

Delivery

- The complete expulsion or extraction from a woman of one or more fetuses, after at least 22 completed weeks of gestational age, irrespective of whether they are live births or stillbirths. A delivery of either a single or multiple newborn is considered as one delivery. If more than one newborn is delivered, it is often recognized as a delivery with multiple births.
- The expulsion or extraction of one or more fetuses from the mother after 20 completed weeks of gestational age.

Delivery rate

- The number of deliveries expressed per 100 initiated cycles, aspiration cycles, or embryo transfer cycles. When delivery rates are given, the denominator (initiated, aspirated, or embryo transfer cycles) must be specified. It includes deliveries that resulted in the birth of one or more live babies and/or stillborn babies. Note: The delivery of a singleton, twin, or other multiple pregnancy is registered as one delivery.

- The number of deliveries expressed per 100 initiated cycles, aspiration cycles, or embryo transfer cycles. When delivery rates are recorded, the denominator (initiated, aspirated or embryo transfer cycles) must be specified. It includes deliveries that resulted in the birth of one or more live births and/or stillbirths. The delivery of a singleton, twin or other multiple pregnancy is registered as one delivery. If more than one newborn is delivered, it is often recognized as a delivery with multiple births.

Delivery rate after ART treatment per patient

- The number of deliveries with at least one live born baby per patient after a specified number of ART treatments.

Delivery rate after fertility treatment per patient

- The number of deliveries with at least one live birth or stillbirth, expressed per 100 patients, after a specified time and following all treatments.

Delivery with multiple births after fertility treatments

- A single delivery with more than one newborn, following all fertility treatments.

deoxyribonucleic acid

- DNA; the molecule containing genetic information, composed of a double-stranded helix of nucleotides held together by hydrogen bonds between adenine-thymine and guanine-cytosine pairs.

depth of coverage

- The number of times a nucleotide is read during sequencing, which helps differentiate sequencing errors from true variants.

Diandric oocytes

- An oocyte with an extra set of haploid chromosomes of paternal origin.

Digynic oocytes

- An oocyte with an extra set of haploid chromosomes of maternal origin.

Diminished ovarian reserve

- A term generally used to indicate a reduced number and/or reduced quality of oocytes, such that the ability to reproduce is decreased. (See ovarian reserve.)
- This diagnosis means that the ability of the ovary to produce eggs is reduced. Reasons include congenital, medical, or surgical causes.

Diploid

- A cell with two copies of each chromosome—one from each parent. This is the normal state, except when cells are preparing to divide.

Diploidy/euploidy

- The condition in which a cell has two haploid sets of chromosomes. Each chromosome in one set is paired with its counterpart in the other set. A diploid embryo has 22 pairs of autosomes and two sex chromosomes, the normal condition.

Directed mutation

- See Adaptive mutation.

disease-causing mutation

- A genetic alteration that increases an individual's risk of developing a disease; also known as deleterious mutation, pathogenic variant, or predisposing mutation.

Disomy

- The normal number of chromosomes characterized by 22 pairs of autosomal chromosomes and one pair of sex chromosomes (XX or XY). The chromosome number in human cells is normally 46.

DNA

- Deoxyribonucleic acid (DNA) contains the genetic instructions in all living things. DNA is made up of two strands that wind around each other and looks like a twisting ladder (a shape called a double helix). A DNA strand has four different bases arranged in different orders. These bases are T (thymine), A (adenine), C (cytosine), and G (guanine). DNA is "read" by the order of the bases, that is by the order of the Ts, Cs, Gs, and As. The specific order, or sequence, of these bases determines the exact information carried in each gene (for example, instructions for making a specific protein). DNA has the same structure in every gene and in almost all living things.
- DeoxyriboNucleic Acid, the genetic material consisting of a long chain of individual units called nucleotides, each consisting of a base joined to a sugar and a phosphate group.
- Deoxyribonucleic acid, the molecule that contains genetic information. Also called deoxyribonucleic acid.
- Deoxyribonucleic acid, the molecule that conveys genetic information about protein sequence and expression across generations. Discovered in 1869, its significance was widely recognized starting in the 1950s.

DNA Methylation

- DNA methylation is a chemical addition to a piece of DNA that turns it on or off.

DNA methylation

- An endogenous process in the cell which adds a methyl group, -CH3, to the base cytosine or adenosine, resulting in gene-silencing, or failure of the gene to become expressed.

DNA Mutation

- A mutation is a change in a DNA sequence. DNA mutations in a gene can change what protein is made. Mutations present in the eggs and sperm (germline mutations) can be passed on from parent to child, while mutations that occur in body cells (somatic mutations) cannot be inherited.

DNA polymerase

- An enzyme that makes DNA.

DNA sequence

- The sequence of bases in a stretch of DNA. There are 4 different bases, which are simply represented by the alphabets, A, T, C and G. An example of a DNA sequence is as follows: ATTTCCGCTACGCGTTA… An RNA sequence is similar, except that the alphabet T is replaced by U.

DNAse

- Deoxyribonuclease, an enzyme that breaks down DNA.

domain

- A specific physical region or amino acid sequence in a protein associated with a particular function, or the corresponding segment in DNA.

Dominant

- A Mendelian trait that is expressed when only one copy of the allele is present. Such traits appear even if only one parent contributes the allele.
- Dominant diseases can be caused by only one copy of a gene with a DNA mutation. If one parent has a disease, each child has a 50% chance of inheriting the mutated gene.

Dominant allele

- An allele which is expressed when only one copy is present in an individual, i.e., in heterozygous condition.

Donated embryo cycle

- An ART cycle where an embryo is donated by a patient or couple who previously underwent ART treatment and had extra embryos available and then transferred to another woman (the recipient).

Donor egg cycle

- An ART cycle where an embryo is formed from the egg of one woman (the donor) and then transferred to another woman (the recipient). Sperm from either the recipient's partner or a donor may be used.

Donor insemination

- The process of placing laboratory processed sperm or semen from a man into the reproductive tract of a woman who is not his intimate sexual partner, for the purpose of initiating a pregnancy.

Double embryo transfer (DET)

- The transfer of two embryos in an ART procedure. This may be elective (eDET) when more than two embryos of sufficient quality for transfer are available.

double heterozygosity

- The presence of two different mutated alleles at two separate genetic loci.

D'Amico criteria

- A set of clinical criteria used to stratify prostate cancers into risk categories based on Gleason score, PSA level, and clinical stage.

each-we dilemma

- A term philosophers, as used by Derek Parfit, to designate choices in which what is best for each of us is not best for all of us (see collective action problem).

Early neonatal death

- Death of a live born baby within 7 days of birth.

Early neonatal death/mortality

- Death of a newborn within 7 days of birth.

Easy PGD

- A term coined to describe inexpensive whole genome analysis of an embryo before implantation when the egg that contributed to the embryo was artificially derived from stem cells.

Ecosystem

- The totality of all plant and animal species that constitute an interdependent, interrelated community.

Ectopic pregnancy

- A pregnancy in the fallopian tube or elsewhere outside the lining of the uterus.
- A pregnancy in which implantation takes place outside the uterine cavity. (More? Ectopic Implantation)
- A pregnancy outside the uterine cavity, diagnosed by ultrasound, surgical visualization or histopathology.
- A pregnancy where the fertilized egg implants in a location outside of the uterus—usually in the fallopian tube, the ovary, or the abdominal cavity. Ectopic pregnancy is a dangerous condition that requires prompt medical treatment.

Egg

- A female reproductive cell, also called an oocyte or ovum.

Egg (oocyte)

- The female sex cell (ovum) produced by the ovary, which, when fertilized by a male's sperm, produces an embryo.

Egg or Embryo banking cycle

- An ART cycle started with the intent of freezing (cryopreserving) all resulting eggs or embryos for potential future use. This may include freezing with the intent for future use in the short term (12 months) or long term (>12 months). Long-term egg or embryo banking is also called fertility preservation.

Egg retrieval

- The procedure in which eggs are obtained by inserting a needle into the ovarian follicle and removing the fluid and the egg by suction. Also called oocyte aspiration.

Egg retrieval (also called oocyte retrieval)

- A procedure to collect the eggs contained in the ovarian follicles.

Ejaculation

- Co-ordinated contractions of the genitourinary tract leading to the ejection of spermatozoa and seminal fluid.

Ejaculation retardata

- A condition resulting in an inability to ejaculate during vaginal intercourse.

Ejaculatory duct

- The canal that passes through the prostate just lateral to the verumontanum where the vas deferens and the duct from the seminal vesicle coalesce.

Elective embryo transfer

- The transfer of one or more embryos, selected from a larger cohort of available embryos.

Elective single embryo transfer (eSET)

- The transfer of one (a single) embryo selected from a larger cohort of available embryos.

Electroejaculation (EEJ)

- Procedure to cause ejaculation of sperm, performed by electrical stimulation of tissue in the region of the prostate.

Embryo

- A fertilized egg that has begun cell division.
- An egg that has been fertilized by a sperm and has then undergone one or more cell divisions.
- The biological organism resulting from the development of the zygote, until eight completed weeks after fertilization, equivalent to 10 weeks of gestational age.
- The earliest developmental stage of a multicellular eukaryote. In humans, it generally begins at fertilization and lasts until the end of the eighth week, when it is renamed a fetus. Different early stages may also be called blastocyst or morula.
- The product of the division of the zygote to the end of the embryonic stage, 8 weeks after fertilization. (This definition does not include either parthenotes—generated through parthenogenesis—nor products of somatic cell nuclear transfer.)

Embryo bank

- Repository of cryopreserved embryos stored for future use.

Embryo culture

- Growth of the embryo in a laboratory (culture) dish.

Embryo donation

- The transfer of an embryo resulting from gametes (spermatozoa and oocytes) that did not originate from the recipient and her partner.

Embryo donation (for reproduction)

- An ART cycle, which consists of the transfer of an embryo to the uterus or Fallopian tube of a female recipient, resulting from gametes that did not originate from the female recipient or from her male partner, if present.

Embryo fragmentation

- The process during which one or more blastomeres shed membrane vesicles containing cytoplasm and occasionally whole chromosomes or chromatin.

Embryo recipient cycle

- An ART cycle in which a woman receives zygote(s) or embryo(s) from donor(s).
- An ART cycle in which a woman's uterus is prepared to receive one or more cleavage stage embryos/blastocysts, resulting from gametes that did not originate from her or from her male partner, if present.

Embryo transfer

- (ET) The procedure in which one or more embryos are placed in the uterus or fallopian tube.
- Placement of an embryo into the uterus or, in the case of ZIFT and TET, into the fallopian tube.
- Placement of embryos into a woman's uterus through the cervix after IVF.

Embryo transfer (ET)

- Placement into the uterus of an embryo at any embryonic stage from day 1 to day 7 after IVF or ICSI. Embryos from day 1 to day three can also be transferred into the Fallopian tube.

Embryo transfer cycle

- An ART cycle in which one or more embryos are transferred into the uterus or fallopian tube.
- An ART cycle in which one or more fresh or frozen/thawed embryos at cleavage or blastocyst stage are transferred into the uterus or Fallopian tube.

Embryo/fetus reduction

- A procedure to reduce the number of viable embryos or fetuses in a multiple pregnancy.

Emission (semen)

- Co-ordinated contractions of the vas deferentia, seminal vesicles, and ejaculatory ducts leading to deposition of semen into the urethral meatus prior to ejaculation.

Empty follicle syndrome

- (EFS) Clinically defined as the failure to aspirate oocytes from mature ovarian follicles during in vitro fertilization. At least one genetic cause has been identified, a missense mutation in zona pellucida protein 3 (ZP3).[3]

Endometriosis

- A disease characterized by the presence of endometrium-like epithelium and stroma outside the endometrium and myometrium. Intrapelvic endometriosis can be located superficially on the peritoneum (peritoneal endometriosis), can extend 5 mm or more beneath the peritoneum (deep endometriosis) or can be present as an ovarian endometriotic cyst (endometrioma).
- A disease in which tissue resembling endometrium (the lining of the uterus) grows outside the uterus. It is often associated with infertility.
- A medical condition that involves the presence of tissue similar to the uterine lining in locations outside the uterus, such as the ovaries, fallopian tubes, or abdominal cavity.

Entropy

- A measure of the disordered, degraded energy that is unavailable for work.

Environmental Factors

- Environmental factors can include exposures related to where we live as well as behaviors such as smoking and exercise and cultural factors such as foods that we eat.

Enzyme

- A Protein produced by living organisms that acts as a catalyst for a specific biochemical (metabolic) reaction.

Epididymis

- A convoluted, highly coiled duct that transports the spermatozoa from the testis via the efferent ducts to the vas deferens.
- The duct between testes and vas deferens where sperm are stored and mature.

epigenetic alteration

- A heritable change in gene expression that does not involve changes to the DNA sequence, such as promoter methylation or histone modification; also called epigenetic variant or epimutation.

epigenetic variant

- A heritable change in gene expression without altering the DNA sequence, such as promoter methylation; also called epigenetic alteration or epimutation.

Epigenetics

- Epigenetics is the study of changes in phenotype caused by something other than changes in the underlying DNA sequence (for example, DNA methylation).

epigenetics

- The study of heritable changes in gene expression that do not involve alterations to the underlying DNA sequence.

epimutation

- A heritable change in gene expression that does not involve changes to the DNA sequence, such as promoter methylation or histone modifications; also called epigenetic alteration or variant.

Epistasis

- Interaction between genes.

ER-negative PR-negative HER2/neu-negative breast cancer

- A breast cancer defined by a lack of expression of estrogen receptor, progesterone receptor, and HER2/neu; also called triple-negative breast cancer (TNBC).

Erectile dysfunction

- Inability to have and/or sustain an erection sufficient for intercourse.

Estradiol

- The predominant estrogen (hormone) produced by the follicular cells of the ovary.

Estrogen

- The female hormone largely responsible for thickening the uterine lining during the first half of the menstrual cycle in preparation for ovulation and possible pregnancy. Estradiol is the main estrogen.

eugenics

- The belief that we should use the science of genetics to try to improve the welfare of our children. The word was coined by Galton from the Greek roots eu + gen (good + birth) to indicate the study of genetics in the service of creating future people.

Eukaryote

- An organism whose cells contain a nucleus and other membrane-bound organelles. Eukaryotes include multicellular organisms and some single-celled organisms, as opposed to prokaryotes (bacteria and archaea).
- The major class of living things, including all multicellular, higher organisms and some single-celled organisms, that have a nucleus in their cells, containing the chromosomes.

Euploid

- A cell with the correct number of chromosomes. In humans, this is usually 46 chromosomes (22 pairs of autosomes and 2 sex chromosomes), although it can also refer to cells in division or gametes with one set of chromosomes.

Euploidy

- The condition in which a cell has chromosomes in an exact multiple of the haploid number; in the human this multiple is normally two. Thus, a normal embryo that is euploid is also diploid.

Excessive ovarian response

- An exaggerated response to ovarian stimulation characterized by the presence of more follicles than intended. Generally, more than 20 follicles >12 mm in size and/or more than 20 oocytes collected following ovarian stimulation are considered excessive, but these numbers are adaptable according to ethnic and other variables.

Exome

- The portion of the genome made up of exons, the regions of DNA that specify the order and identity of amino acids in proteins. It represents less than 2% of the human genome.

exon

- The portion of a gene that is present in the final, mature messenger RNA transcript; most exons code for amino acids.

Exon

- The part of a gene that specifies the order and identity of amino acids in a protein. In eukaryotes, exons are separated by introns.

Expectant fertility management

- Management of fertility problems including infertility without any specific active clinical or therapeutic interventions other than fertility information and advice, to improve natural fertility, based upon the probability of becoming pregnant.

externality

- A cost or benefit borne by someone external to a transaction.

Extremely low birth weight

- Birth weight less than 1,000 g. (More? Birth-Weight)
- Birth weight less than 1000 g.

Extremely preterm birth

- A birth that takes place after 22 but before 28 completed weeks of gestational age.
- A live birth or stillbirth that takes place after 20 but before 28 completed weeks of gestational age.

facies

- A distinctive facial appearance or expression characteristic of a specific condition.

Fallopian tubes

- A pair of tubes attached to the uterus, one on each side, where sperm and egg meet in normal conception.

false-positive result

- A test result that incorrectly indicates the presence of a gene mutation or condition in an unaffected individual.

familial

- Relating to a trait or condition that occurs more frequently in a specific family than in the general population.

family cancer syndrome

- An inherited disorder with a higher-than-normal risk of certain cancers, such as Lynch syndrome or hereditary breast and ovarian cancer syndrome; also called hereditary or inherited cancer syndrome.

FDA

- The Food and Drug Administration, the US agency responsible for regulating drugs, medical devices, and biological products, among other items.

FDCA

- The Federal Food, Drug, and Cosmetic Act, passed in 1938 and frequently amended, which gives the FDA its regulatory power.

FDR

- A first-degree relative; a parent, sibling, or child of an individual.

Fecundability

- The probability of a pregnancy, during a single menstrual cycle in a woman with adequate exposure to sperm and no contraception, culminating in a live birth. In population-based studies, fecundability is frequently measured as the monthly probability.

Fecundity

- Clinically defined as the capacity to have a live birth.

Female infertility

- Infertility caused primarily by female factors encompassing: ovulatory disturbances; diminished ovarian reserve; anatomical, endocrine, genetic, functional or immunological abnormalities of the reproductive system; chronic illness; and sexual conditions incompatible with coitus.

Fertility

- The capacity to establish a clinical pregnancy.

Fertility awareness

- The understanding of reproduction, fecundity, fecundability, and related individual risk factors (e.g. advanced age, sexual health factors such as sexually transmitted infections, and lifestyle factors such as smoking, obesity) and non-individual risk factors (e.g. environmental and workplace factors); including the awareness of societal and cultural factors affecting options to meet reproductive family planning, as well as family building needs.

Fertility care

- Interventions that include fertility awareness, support and fertility management with an intention to assist individuals and couples to realize their desires associated with reproduction and/or to build a family.

Fertility preservation

- Various interventions, procedures and technologies, including cryopreservation of gametes, embryos or ovarian and testicular tissue to preserve reproductive capacity.

Fertility preservation cycle

- An ART cycle started with the intent of freezing and banking all eggs or embryos for more than 12 months for future use.

Fertilization

- (fertilisation) The penetration of the ovum by the spermatozoon and combination of their genetic material resulting in the formation of a zygote. (More? Fertilization)
- A sequence of biological processes initiated by entry of a spermatozoon into a mature oocyte followed by formation of the pronuclei.
- The fusion of sperm and egg.
- The penetration of the egg by the sperm and the resulting combination of genetic material that may develop into an embryo.

FET

- See Frozen-thawed embryo transfer cycle.

Fetal death (stillbirth)

- Death before the complete expulsion or extraction from its mother of a product of fertilization, at or after 20 completed weeks of gestational age. The death is indicated by the fact that, after such separation, the fetus does not breathe or show any other evidence of life, such as heart beat, umbilical cord pulsation, or definite movement of voluntary muscles.

Fetal loss

- Death of a fetus. It is referred to as early fetal loss when death takes place between 10 and 22 weeks of gestational age; late fetal loss, when death takes place between 22 and 28 weeks of gestational age; and stillbirth when death takes place after 28 weeks gestational age.

Fetus

- The product of fertilization from completion of embryonic development, at 8 completed weeks after fertilization, until abortion or birth.
- The stages of development of an organism from eight completed weeks of fertilization (equivalent to 10 weeks of gestational age) until the end of pregnancy

fibrofolliculoma

- A benign tumor of the hair follicle that appears as a small, whitish papule; pathognomonic for Birt-Hogg-Dubé syndrome.

Fibroids

- Benign (non-cancerous) tumors of the uterine muscle wall that can cause abnormal uterine bleeding and pain.

first-degree relative

- A parent, sibling, or child of an individual; also called FDR.

FISH

- Fluorescence in situ hybridization, a technique that uses fluorescent markers attached to DNA or RNA probes to detect complementary sequences in chromosomes.
- Fluorescence in situ hybridization; a technique that uses fluorescently labeled DNA probes to detect specific chromosomes or chromosomal regions.

fluorescence in situ hybridization

- A technique using fluorescently labeled DNA probes to detect specific chromosomes or regions, also called FISH.

Follicle

- A fluid-filled structure in the ovary containing an egg and the surrounding cells that produce hormones. As the follicle matures, the fluid can be visualized by ultrasound.
- A structure in the ovaries that contains a developing egg.

Follicle-stimulating hormone (FSH)

- The pituitary hormone responsible for stimulating the growth of the follicle that surrounds the egg. In addition, it is the hormone in injectable ovulation medications that promotes growth of the follicles.

founder mutation

- A genetic alteration observed at high frequency in an isolated group, resulting from an ancestor being a carrier; also called founder variant.

founder variant

- A genetic alteration observed at high frequency in an isolated group, resulting from an ancestor being a carrier; also called founder mutation.

frameshift mutation

- An insertion or deletion of base pairs not in multiples of three, disrupting the reading frame and often resulting in a premature stop codon and truncated protein; also called frameshift variant.

frameshift variant

- An insertion or deletion of base pairs not in multiples of three, disrupting the reading frame and often leading to a premature stop codon and truncated protein; also called frameshift mutation.

Freeze-all cycle

- An ART cycle in which, after oocyte aspiration, all oocytes and/or embryos are cryopreserved and no oocytes and/or embryos are transferred to a woman in that cycle.

Fresh eggs, sperm, or embryos

- Eggs, sperm, or embryos that have not been frozen.

Fresh embryo cycle

- An ART cycle where fresh (never frozen) embryos are transferred to the woman. Fresh or frozen eggs may be combined with fresh or frozen sperm to create fresh embryos.

Frozen embryo cycle

- An ART cycle where frozen (cryopreserved) embryos are thawed and transferred to the woman. Fresh or frozen eggs may be combined with fresh or frozen sperm to create frozen embryos.

Frozen-thawed embryo transfer (FET) cycle

- An ART procedure in which cycle monitoring is carried out with the intention of transferring to a woman, frozen/thawed or vitrified/warmed embryo(s)/blastocyst(s). Note: A FET cycle is initiated when specific medication is provided or cycle monitoring is started in the female recipient with the intention to transfer an embryo.

Frozen-thawed embryo transfer cycle

- (FET) an ART procedure in which cycle monitoring is carried out with the intention of transfering frozen-thawed embryo(s). Note: An FET cycle is initiated when specific medication is provided or cycle monitoring is started with the intention to treat.

Frozen-thawed oocyte cycle

- An ART procedure in which cycle monitoring is carried out with the intention of fertilizing thawed oocytes and performing embryo transfer.
- An ART procedure in which cycle monitoring is carried out with the intention of fertilizing thawed/warmed oocytes and performing an embryo transfer.

Full-term birth

- A birth that takes place between 37 and 42 completed weeks of gestational age.
- A live birth or stillbirth that takes place between 37 and 42 completed weeks of gestational age.

game

- A situation in which the outcome for the relevant players is a function of the choices of each player. For example, in poker, winning depends not just on my cards but also on the cards everyone else has.

game theory

- The study of games, including the strategies most likely to work for a particular player, and the outcomes most likely to be stable. For example, nuclear deterrence can achieve peace when each party believes the other will retaliate to a first strike.

Gamete

- A mature germ cell (egg or sperm) that can unite with a gamete of the opposite sex to form a zygote.

Gamete intrafallopian transfer

- (GIFT) An ART procedure in which both gametes (oocytes and spermatozoa) are transferred to the fallopian tube.

Gamete intrafallopian transfer (GIFT)

- An ART procedure in which both gametes (oocytes and spermatozoa) are transferred into a Fallopian tube(s).
- The direct transfer of sperm and eggs into the fallopian tube. Fertilization takes place inside the tube.

Gene

- A gene is a part of DNA that carries the information needed to make a protein. People inherit one copy of each gene from their mother and one copy from their father. The genes that a person inherits from his or her parents can determine many things. For example, genes affect what a person will look like and whether the person might have certain diseases.
- A unit of heredity embodied in DNA that specifies the structure for a protein or a functional RNA molecule (excluding messenger or transfer RNA).
- A unit of heredity, usually a stretch of genetic material (DNA or RNA) with a defined function in the organism or cell, such as one for a protein. There are many genes within a genome. For example, the human genome is now found to contain about 30 000 genes, while the rice genome has about 50 000.

gene

- The basic unit of heredity, made up of DNA sequences that encode proteins or functional RNA, located on chromosomes.

Gene amplification

- The process whereby genes or a sequence of DNA in the genome is greatly increased in number of copies.

Gene cloning

- The technique of making many copies of a gene, isolating the gene and identifying it.

gene editing

- Altering specific variants of DNA in order to achieve a phenotypic effect. CRISPR is currently the most powerful and accurate tool for editing genes.

Gene expression

- The synthesis of the gene-product or protein encoded by the gene.

Gene Expression

- Gene expression refers to the process of making proteins using the instructions from genes. Changes in gene expression can affect how much of a protein is made, as well as when the protein is made.

Gene family

- A group of very similar genes, usually produced by duplication of an original gene, that often perform similar functions.

Gene silencing

- The process(es) whereby certain genes in the genome are prevented from being expressed by chemical modifications and other means.

Gene therapy

- Treating diseases by replacing the defective gene, either by incorporating a normal copy of the gene in the germ-cells (egg or sperm) or in the embryo (germline gene replacement therapy), or by supplying copies of the normal gene to be taken up and incorporated into cells of the adult (somatic cell gene replacement therapy).

genetic anticipation

- A phenomenon where the signs and symptoms of a genetic condition become more severe or appear at an earlier age in successive generations.

Genetic code

- The code establishing the correspondence between the sequence of bases in nucleic acids (DNA and the complementary RNA) and the sequence of amino acids in proteins.

genetic counseling

- A process to help individuals and families understand the risks, implications, and management options for genetic disorders.

Genetic determinism

- Determinism is the doctrine that all acts, choices and events are the inevitable consequence of antecedent sufficient causes. Genetic determinism is the doctrine that the organism is the inevitable consequence of its genetic makeup, or the sum of its genes.

Genetic engineering

- The manipulating of genetic material in the laboratory. It includes isolating, copying, and multiplying genes, recombining genes or DNA from different species, and transferring genes from one species to another, bypassing the reproductive process.

genetic enhancement

- Any genetic intervention that is expected to increase the chances of a person leading a good life. On this definition, calling something an enhancement does not commit us to saying it would be good, all things considered.

genetic heterogeneity

- Different genetic mechanisms (allelic or locus heterogeneity) that produce the same or similar phenotypes.

genetic marker

- A DNA sequence with a known location on a chromosome used as a reference point for mapping genes.

Genetic marker

- Any segment of DNA that can be identified, or whose chromosomal location is known, so that it can be used as a reference point to map or locate other genes. Any gene which has an identifiable phenotype that can be used to track the presence or absence of other genes on the same piece of DNA transferred into a cell.

Genetic modification or transgenesis

- The process whereby a genetically modified organism is made in the laboratory. This involves making artificial or modified genetic material (GM constructs) which are inserted into the genomes of cells or embryos. The cell or embryo is regenerated to an organism, out of which a GM line or transgenic line is derived.

genetic predisposition

- An increased likelihood of developing a disease based on genetic variants and/or family history; also called genetic susceptibility or inherited predisposition.

genetic screening

- Testing a population to identify individuals at increased risk for a particular genetic disorder or for carrying a specific genetic variant.

genetic susceptibility

- An increased likelihood of developing a disease based on genetic factors; also called genetic predisposition.

Genetically modified organism (GMO)

- An organism which has foreign DNA inserted into its genome by means of genetic modification in the laboratory.

Genetics

- The study of genes and heredity.

genodermatosis

- An inherited syndrome that includes skin abnormalities as part of its phenotype.

Genome

- The complete sequence of DNA in an organism or individual.
- The totality of all the genetic material (deoxyribonucleic acid or DNA) in an organism, organised in a precise, though by no means fixed or constant way. In the case of viruses, most of them will have ribonucleic acid or RNA as the genetic material.

genome-wide association study

- A method to identify genetic variants across the entire genome associated with a disease or trait; also called GWAS.

genomic imprinting

- An epigenetic process in which the expression of an allele depends on the parent of origin.

Genomics

- Genomics refers to the study of all of the genetic material in an organism.
- The study of large-scale DNA sequences and genome structure, rather than individual genes.

Genotype

- The genetic makeup of a cell or organism, often referring to specific DNA variations present.
- The genotype of a person is her or his genetic makeup. It can also refer to the alleles that a person has for a specific gene.
- The precise variant(s) of the gene(s) carried by an individual.

genotype

- The genetic makeup of an individual at a specific locus or for the entire genome.

genotyping

- The laboratory process of analyzing an individual's DNA to determine the presence of specific genetic variants.

Germinal vesicle (GV)

- The nucleus in an oocyte at prophase I.

germline

- Cells from which eggs or sperm are derived.

germline DNA

- DNA derived from reproductive cells that is incorporated into every cell of the offspring; also called constitutional DNA.

germline mosaicism

- The presence of a pathogenic variant confined to the eggs or sperm, such that a parent may pass it on without showing symptoms; also called gonadal mosaicism.

germline mutation

- A mutation in a reproductive cell that is present in every cell of the offspring and is heritable; also called germline variant.

germline variant

- A variant in a reproductive cell that is present in every cell of the offspring and is heritable; also called germline mutation.

Gestational age

- Age of an embryo or fetus calculated by adding 2 weeks (14 days) to the number of completed weeks since fertilization. Note: For frozen-thawed embryo transfers, an estimated date of fertilization is computed by subtracting the embryo age at freezing from the transfer date of the FET cycle.
- The age of an embryo or fetus calculated by the best obstetric estimate determined by assessments which may include early ultrasound and the date of the last menstrual period and/or perinatal details. In the case of ART, it is calculated by adding two weeks (14 days) to the number of completed weeks since fertilization. Note: For frozen-thawed embryo transfer (FET) cycles, an estimated date of fertilization is computed by subtracting the combined number of days an embryo was in culture pre-cryopreservation and post-thaw/-warm, from the transfer date of the FET cycle.
- The time from estimated last menstrual period (LMP) to birth. LMP for ART cycles is estimated using the date of retrieval or transfer.

Gestational carrier

- (surrogate) A woman who carries a pregnancy with an agreement that she will give the offspring to the intended parent(s). Gametes can originate from the intended parent(s) and/or a third party (or parties).
- A woman who carries a pregnancy for another couple. The pregnancy is derived from the egg and sperm of the couple. Although she carries the pregnancy to term, she does not have a genetic relationship to the resulting child.
- A woman who carries a pregnancy with an agreement that she will give the offspring to the intended parent(s). Gametes can originate from the intended parent(s) and/or a third party (or parties). This replaces the term 'surrogate.'

Gestational carrier (also called a gestational surrogate)

- A woman who gestates, or carries, an embryo that was formed from the egg of another woman with the expectation of returning the infant to its intended parents.

Gestational sac

- A fluid-filled structure associated with early pregnancy, which may be located inside or outside the uterus (in case of an ectopic pregnancy).
- A fluid-filled structure associated with early pregnancy, which may be located inside or, in the case of an ectopic pregnancy, outside the uterus.

GIFT

- See Gamete intrafallopian transfer.

GIFT (gamete intrafallopian transfer)

- An ART procedure that involves removing eggs from the woman's ovary and using a laparoscope to place the unfertilized gametes (eggs and sperm) into the woman's fallopian tube through small incisions in her abdomen.

Globozoospermia

- Describes spermatozoa with a reduced or absent acrosome.

GnRH agonists

- A GnRH analog that initially stimulates the pituitary gland to release LH and FSH, followed by a delayed suppressive effect. GnRH agonists are also used to help stimulate follicle growth when started at the beginning of an IVF cycle.

GnRH analogs

- Synthetic hormones similar to the naturally occurring gonadotropin releasing hormone used to prevent premature ovulation. There are two types of GnRH analogs: GnRH agonists and GnRH antagonists.

GnRH antagonists

- Synthetic hormones similar to the naturally occurring gonadotropin releasing hormone used to prevent premature ovulation. These medications have an immediate suppressive effect on the pituitary gland.

gonadal mosaicism

- The presence of a pathogenic variant confined to the ovaries or testes, where a parent may pass it on without being affected; also called germline mosaicism.

Gonadotropin releasing hormone (GnRH)

- Hormone secreted by the hypothalamus, a control center in the brain, which prompts the pituitary gland to release FSH and LH into the bloodstream.

GWAS

- A genome-wide association study, which examines many DNA variations across the genome to determine associations with a given trait.
- Genome-wide association study; a method to identify genetic variants associated with a disease or trait by scanning the entire genome.

Haploid

- A cell with only half the normal number of chromosomes, as produced by meiosis. In humans, haploid cells have 23 chromosomes.

Haploidy

- The condition in which a cell has one set of each of the 23 single chromosomes. Mature human gametes are haploid, each having 23 single chromosomes.

haploinsufficiency

- The situation that occurs when one copy of a gene is inactivated or deleted and the remaining functional copy of the gene is not adequate to produce the needed gene product to preserve normal function.

haplotype

- A set of closely linked genetic markers or DNA variations on a chromosome that tend to be inherited together.

Hatching

- The process by which an embryo at the blastocyst stage extrudes out of, and ultimately separates from, the zona pellucida.
- The process by which an embryo at the blastocyst stage separates from the zona pellucida. (More? Blastocyst Development(

hemizygous

- Describes an individual who has only one member of a chromosome pair or chromosome segment rather than the usual two. Hemizygosity is often used to describe X-linked genes in males who have only one X chromosome. This term is sometimes used in somatic cell genetics where cancer cell lines are often hemizygous for certain alleles or chromosomal regions.

hereditary cancer syndrome

- A type of inherited disorder in which there is a higher-than-normal risk of certain types of cancer. Hereditary cancer syndromes are caused by inherited genetic variants and may be associated with several clinical manifestations. Examples include Lynch syndrome and hereditary breast and ovarian cancer syndrome. Also called family cancer syndrome and inherited cancer syndrome.

hereditary predisposition

- An increased chance or likelihood of developing a particular disease based on the presence of one or more genetic variants and/or a family history suggestive of an increased risk of the disease. Having a hereditary predisposition does not mean an individual will develop the disease. Also called genetic predisposition, genetic susceptibility, and inherited predisposition.

heritability

- A metric behavior geneticists use to measure how much genes explain the differences between individuals within a given population.
- The proportion of variation in a population trait that can be attributed to inherited genetic factors. Heritability estimates range from 0 to 1 and are often expressed as a percentage. A number close to 1 may indicate a highly heritable trait, but it should not be used to estimate individual risk.

hESCs

- Human embryonic stem cells derived from the inner cell mass of blastocysts, maintained in an undifferentiated state in culture.

Heterochromatin

- Regions of tightly packed DNA in chromosomes that are rarely expressed and difficult to sequence, contrasted with euchromatin which contains most active genes.

heteroduplex analysis

- A laboratory method used to detect sequence differences between normal DNA and the DNA to be tested. It is commonly used to screen for point mutations but does not pinpoint the exact location of the variant.

heterogeneity logarithm of the odds score

- A statistical estimate of whether two genetic loci are close enough on a chromosome to be inherited together. It is calculated in the presence of locus heterogeneity and is also called HLOD score.

Heterotopic pregnancy

- Concurrent pregnancy involving at least one embryo implanted in the uterine cavity and at least one implanted outside of the uterine cavity.

Heterozygote

- An individual who has two different alleles of a gene.

heterozygous genotype

- The presence of two different alleles at a particular gene locus. A heterozygous genotype may include one normal allele and one mutated allele or two different mutated alleles (compound heterozygote).

High-order multiple

- A pregnancy or delivery with three or more fetuses or neonates.

High-order multiple births

- The complete expulsion or extraction from their mother of three or more fetuses, after 22 completed weeks of gestational age, irrespective of whether they are live births or stillbirths.

High-order multiple gestation

- A pregnancy with three or more embryos or fetuses.

HLOD score

- A statistical estimate of whether two genetic loci are near enough on a chromosome to be inherited together, calculated in the presence of locus heterogeneity. Also called heterogeneity logarithm of the odds score.

Homozygote

- An individual who has two identical alleles of the gene.

homozygous genotype

- The presence of two identical alleles at a particular gene locus. A homozygous genotype may include two normal alleles or two alleles with the same mutation.

Horizontal gene transfer

- The direct uptake of foreign genetic material by cells and incorporation into the cells' genome.

HTC/P

- Human cells, tissues, and cellular or tissue-based products, a regulatory classification used by the FDA.

Human chorionic gonadotropin (hCG)

- A hormone produced by the placenta; its detection is the basis for most pregnancy tests. Also refers to the medication used to induce ovulation and during the final stages of egg maturation.

Human menopausal gonadotropin (hMG)

- An ovulation drug that contains follicle stimulating hormone (FSH) and luteinizing hormone (LH) derived from the urine of postmenopausal women. hMG is used to stimulate the growth of multiple follicles.

Hydrosalpinx

- A blocked, dilated, fluid-filled fallopian tube.
- A distally occluded, dilated, fluid-filled Fallopian tube.

Hypergonadotropic hypogonadism

- Gonadal failure associated with reduced gametogenesis, reduced gonadal steroid production and elevated gonadotropin production.

Hyperspermia

- High volume of ejaculate above the upper reference limit. When reporting results, the reference criteria should be specified.

Hypogonadotropic hypogonadism

- Gonadal failure associated with reduced gametogenesis and reduced gonadal steroid production due to reduced gonadotropin production or action.

hypomelanosis

- Deficiency of melanin.

Hypospermatogenesis

- Histopathologic description of reduced production of spermatozoa in the testes.

Hypospermia

- Low volume of ejaculate below the lower reference limit. When reporting results, the reference criteria should be specified.

Iatrogenic testicular failure

- Damage to testicular function after radiation, chemotherapy or hormone treatment; or devascularization as a consequence of hernia surgery.

ICSI

- Intracytoplasmic sperm injection, an assisted reproduction technique where a single sperm is injected directly into an egg.

ICSI (intracytoplasmic sperm injection)

- A procedure where a single sperm is injected directly into an egg; this procedure is commonly used to treat male infertility.

Immature oocyte

- An oocyte at prophase of meiosis I, (i.e. an oocyte at the germinal vesicle (GV)-stage.)

Implantation

- The attachment and subsequent penetration by a zona-free blastocyst into the endometrium, but when it relates to an ectopic pregnancy, into tissue outside the uterine cavity. This process starts 5 to 7 days after fertilization of the oocyte usually resulting in the formation of a gestation sac.
- The attachment and subsequent penetration by the zona-free blastocyst (usually in the endometrium) that starts 5 to 7 days after fertilization.

Implantation rate

- The number of gestational sacs observed divided by the number of embryos transferred (usually expressed as a percentage, %).
- The number of gestational sacs observed divided by the number of embryos transferred.
- When the ART cycle results in an intrauterine clinical pregnancy, the implantation rate is defined as the larger of either the maximum number of fetal hearts detected by ultrasound or the maximum number of infants born, including live-birth deliveries and stillbirths, out of the total number of embryos transferred.

In vitro fertilization

- (IVF) An ART procedure that involves extracorporeal fertilization.

In vitro fertilization (IVF)

- A process in which an egg and sperm are combined in a laboratory dish to facilitate fertilization. If fertilized, the resulting embryo is transferred to the uterus.
- A sequence of procedures that involves extracorporeal fertilization of gametes. It includes conventional in vitro insemination and ICSI.

in vitro fertilization (IVF)

- A procedure that involves combining a sperm and egg outside of the body before implanting it inside the body to induce pregnancy.

In vitro maturation (IVM)

- A sequence of laboratory procedures that enable extracorporeal maturation of immature oocytes into fully mature oocytes that are capable of being fertilized with potential to develop into embryos.

incomplete penetrance

- When some individuals who carry a pathogenic variant do not express the associated phenotype. Also called reduced penetrance.

inconclusive result

- A negative test result in an individual where a clearly deleterious mutation has not been found in any family members; such results must be interpreted in the context of personal and family history. Also called indeterminate or uninformative result.

Indels

- Insertions and deletions in DNA sequences that can cause frameshifts if not divisible by three.

indeterminate result

- A negative test result in an individual where a clearly deleterious mutation has not been found in any family members; also called inconclusive or uninformative result.

Induced abortion

- Intentional loss of an intrauterine pregnancy, through intervention by medical, surgical or unspecified means. (See induced embryo/fetal reduction.)
- The termination of a clinical pregnancy by deliberate interference that takes place before 20 completed weeks of gestational age (18 weeks after fertilization) or, if gestational age is unknown, of an embryo/fetus of less than 400 g.

Induced embryo/fetal reduction

- An intervention intended to reduce the number of gestational sacs or embryos/fetuses in a multiple gestation.

Infertility

- (clinical definition) Disease of the reproductive system defined by the failure to achieve a clinical pregnancy after 12 months or more of regular unprotected sexual intercourse.

- A disease characterized by the failure to establish a clinical pregnancy after 12 months of regular, unprotected sexual intercourse or due to an impairment of a person's capacity to reproduce either as an individual or with his/her partner. Fertility interventions may be initiated in less than 1 year based on medical, sexual and reproductive history, age, physical findings and diagnostic testing. Infertility is a disease, which generates disability as an impairment of function.

Infertility counseling

- A professional intervention with the intention to mitigate the physical, emotional and psychosocial consequences of infertility.

informative

- A test result that definitively reveals the presence or absence of the germline genetic alteration associated with the hereditary disorder being assessed.

informed consent

- A process of information exchange between a clinician and an individual or their proxy to facilitate autonomous, informed decision making regarding genetic testing, including discussion of risks, benefits, limitations, and implications. Also called the consent process.

inherited cancer syndrome

- An inherited disorder characterized by a higher-than-normal risk of certain cancers, caused by inherited genetic variants; also called family cancer syndrome or hereditary cancer syndrome.

inherited predisposition

- An increased likelihood of developing a disease based on genetic variants and/or family history; also called genetic predisposition, genetic susceptibility, and hereditary predisposition.

Initiated cycle

- An ART cycle in which the woman receives specific medication for ovarian stimulation, or monitoring in the case of natural cycles, with the intention to treat, irrespective of whether or not follicular aspiration is attempted.

Initiated medically assisted reproduction cycle (iMAR)

- A cycle in which the woman receives specific medication for ovarian stimulation or in which cycle monitoring is carried out with the intention to treat, irrespective of whether or not insemination is performed, follicular aspiration is attempted in an ovarian stimulation cycle or whether egg(s) or embryo(s) are thawed or transferred in a frozen embryo transfer (FET) cycle.

Inner cell mass

- A group of cells attached to the polar trophectoderm consisting of embryonic stem cells, which have the potential to develop into cells and tissues in the human body, except the placenta or amniotic membranes.
- The cluster of cells inside a blastocyst that eventually form the embryo, fetus, and baby, and are the source of hESCs.

Insemination

- Placement of sperm into the uterus or cervix for producing a pregnancy, or adding sperm to eggs in IVF procedures.

insertion

- A type of genetic change involving the addition of a segment of DNA, which can range from a single base to larger segments.

Intended parent(s)

- A couple or person who seek(s) to reproduce with the assistance of a gestational carrier or traditional gestational carrier.

Interrupted genes

- Genes whose coding sequence is interrupted at intervals by long stretches of non-coding sequences. The coding regions came to be known as exons and the non-coding regions as introns. This structure is now found to be characteristic of most eukaryotic genes. The number and size of introns vary greatly, and they are often much longer than the coding sequences. After transcription, the intron regions are removed, or spliced out from the RNA transcript before it is translated into protein.

Intra-cervical insemination

- A procedure in which laboratory processed sperm are placed in the cervix to attempt a pregnancy.

Intra-uterine insemination

- A procedure in which laboratory processed sperm are placed in the uterus to attempt a pregnancy.

Intra-uterine pregnancy

- A state of reproduction in which an embryo has implanted in the uterus.

Intracytoplasmic sperm injection

- (ICSI) A procedure in which a single spermatozoon is injected into the oocyte cytoplasm.

Intracytoplasmic sperm injection (ICSI)

- A micromanipulation procedure in which a single sperm is injected directly into an egg to attempt fertilization, used with male infertility or couples with prior IVF fertilization failure.
- A procedure in which a single spermatozoon is injected into the oocyte cytoplasm.

intron

- The sequence of DNA between exons that is transcribed into RNA but is removed from the mature messenger RNA transcript.

inversion

- A chromosomal defect in which a segment of a chromosome breaks off and reattaches in the reverse direction.

iPSCs

- Induced pluripotent stem cells, which are adult cells reprogrammed to an embryonic-like state capable of forming many cell types.

iterated embryo selection

- A procedure whereby an embryo is selected for desirable characteristics, then biopsied, transformed into a gamete, and combined with another gamete to produce another embryo. The process could in theory be repeated many times to produce a child with radically altered traits.

IUI (intrauterine insemination)

- A medical procedure that involves placing sperm into a woman's uterus to facilitate fertilization. IUI is not considered an ART procedure because it does not involve the manipulation of eggs.

IVF

- In vitro fertilization, an assisted reproduction technique where egg and sperm are united outside the body to form a zygote and embryo, which is then transferred into the uterus.

- See In vitro fertilization.

IVF (in vitro fertilization)

- An ART procedure that involves removing eggs from a woman's ovaries and fertilizing them outside her body. The resulting embryos are then transferred into a woman's uterus through the cervix. IVF is the most common type of ART.

IVF culture medium

- A special fluid into which sperm, eggs, and embryos are placed when outside the human body.

Karyotype

- The number and appearance of chromosomes in a cell, used in genetic testing to detect abnormalities such as aneuploidies or large chromosomal rearrangements.

kindred

- An extended family.

Laparoscopic ovarian drilling

- A surgical method for inducing ovulation in females with anovulatory or oligo-ovulatory polycystic ovarian syndrome, utilizing either laser or electrosurgery.

Laparoscopy

- A surgical procedure that allows viewing of the internal pelvic organs. During the procedure, a long, narrow, fiber optic instrument, called a laparoscope, is usually inserted through an incision in or below the woman's navel. One or more additional incisions may be made for inserting additional instruments.

Large for gestational age

- A birth weight greater than the 90th centile of the sex-specific birth weight for a given gestational age reference. When reporting outcomes, the reference criteria should be specified. If gestational age is unknown, then the birth weight should be registered.

late or variable onset

- Refers to the age at which a disease phenotype appears in individuals carrying a pathogenic variant, often manifesting later in life or variably over a lifetime.

LD

- Where alleles occur together more often than expected by chance due to physical proximity on a chromosome; also called linkage disequilibrium.

Leukospermia

- A high number of white blood cells in semen above the upper reference limit. When reporting results, the reference criteria should be specified.

Leydig cell

- Type of testicular cell located in the interstitial space between the seminiferous tubules, that secretes testosterone.

LINES

- Long interspersed elements, pieces of DNA copied and inserted into new locations by transposons or retrotransposons, contrasted with SINES.

linkage

- The tendency for genes or DNA segments to be inherited together because of their close physical proximity on the same chromosome.

linkage analysis

- A gene-hunting technique that identifies genetic markers co-inherited with a trait in high-risk families to locate disease-causing genes.

linkage disequilibrium

- The non-random association of alleles at different loci due to their physical proximity on a chromosome; also known as LD.

Live birth

- The complete expulsion or extraction from a woman of a product of fertilization, after 22 completed weeks of gestational age; which, after such separation, breathes or shows any other evidence of life, such as heart beat, umbilical cord pulsation or definite movement of voluntary muscles, irrespective of whether the umbilical cord has been cut or the placenta is attached. A birth weight of 500 grams or more can be used if gestational age is unknown. Live births refer to the individual newborn; for example, a twin delivery represents two live births.
- The complete expulsion or extraction from its mother of a product of fertilization, irrespective of the duration of the pregnancy, which, after such separation, breathes or shows any other evidence of life, such as heart beat, umbilical cord pulsation, or definite movement of voluntary muscles, irrespective of whether the umbilical cord has been cut or the placenta is attached.

Live birth delivery rate

- The number of deliveries that resulted in at least one live birth, expressed per 100 cycle attempts. In the case of ART/MAR interventions, they can be initiated cycles, insemination, aspiration cycles or embryo transfer cycles. When delivery rates are given, the denominator (initiated, inseminated, aspirated or embryo transfer cycles) must be specified.
- The number of deliveries that resulted in at least one live born baby, expressed per 100 initiated cycles, aspiration cycles, or embryo transfer cycles. When delivery rates are given, the denominator (initiated, aspirated, or embryo transfer cycles) must be specified.

Live-birth delivery

- The delivery of one or more infants with at least one born alive.

locus

- The physical location of a specific gene on a chromosome.

locus heterogeneity

- The phenomenon where variants at different gene loci cause the same or similar phenotypic expressions of a disease.

LOD score

- A statistical estimate indicating the likelihood that two genetic loci are linked, with a score of 3 or higher suggesting significant linkage; also called logarithm of the odds score.

logarithm of the odds score

- A statistical measure of genetic linkage, where a score of 3 or higher indicates strong evidence for linkage; also called LOD score.

LOH

- Loss of heterozygosity; the loss of the normal allele at a gene locus in a heterozygous individual, leading to the absence of normal gene function.

loss of heterozygosity

- The loss of the normal allele in a heterozygous gene, often leading to a lack of gene function; also called LOH.

Low birth weight

- Birth weight less than 2,500 g. (More? Birth-Weight)
- Birth weight less than 2500 g.

Luteal phase defect

- A poorly defined abnormality of the endometrium presumably due to abnormally low progesterone secretion or action on the endometrium.

Luteal phase support

- Hormonal supplementation in the luteal phase, usually progesterone.

Luteinizing hormone (LH)

- The pituitary hormone that normally causes ovulation and maturation of the egg.

Major congenital anomaly

- A congenital anomaly that requires surgical repair of a defect, is a visually evident or life-threatening structural or functional defect, or causes death.

Male infertility

- Infertility caused primarily by male factors encompassing: abnormal semen parameters or function; anatomical, endocrine, genetic, functional or immunological abnormalities of the reproductive system; chronic illness; and sexual conditions incompatible with the ability to deposit semen in the vagina.

Male infertility or male factor infertility

- Any cause of infertility due to low sperm count or problems with sperm function that makes it difficult for a sperm to fertilize an egg under normal conditions.

Male-factor infertility

- Infertility caused by a problem in the male; for example the inability to ejaculate or insufficient number of sperm.

massively parallel sequencing

- A high-throughput DNA sequencing method that processes many DNA sequences in parallel; also known as next-generation sequencing (NGS).

Maternal spindle transfer

- (MST) The "maternal spindle" is the group of maternal chromosomes within the egg, which are shaped in a spindle. MST involves removing the spindle from the mother's egg before it is fertilised by the father's sperm. The spindle is then placed into a donor egg with healthy mitochondria (from which the donor's spindle, and therefore her nuclear material, has been removed).
- Transfer of the maternal spindle (including maternal chromosomes) from a patient's oocyte into a donated oocyte in which the maternal spindle with chromosomes has been removed.

Mature oocyte

- An oocyte at metaphase of meiosis II, exhibiting the first polar body and with the ability to become fertilized.

Maturing oocyte

- An oocyte that has progressed from prophase I but has not completed telophase I, thus does not exhibit the first polar body.

Medically assisted reproduction

- (MAR) Reproduction brought about through ovulation induction, controlled ovarian stimulation, ovulation triggering, ART procedures, and intrauterine, intracervical, and intravaginal insemination with semen of husband/partner or donor.

Medically assisted reproduction (MAR)

- Reproduction brought about through various interventions, procedures, surgeries and technologies to treat different forms of fertility impairment and infertility. These include ovulation induction, ovarian stimulation, ovulation triggering, all ART procedures, uterine transplantation and intra-uterine, intracervical and intravaginal insemination with semen of husband/partner or donor.

melanocytic macules associated with Peutz-Jeghers syndrome

- Benign pigmented patches, commonly found on the lips or inside the mouth, that are characteristic of Peutz-Jeghers syndrome, a hereditary condition associated with various cancers.

MESA

- Acronym for microsurgical epididymal sperm aspiration.

MESE

- Acronym for microsurgical epididymal sperm extraction.

Messenger RNA

- The RNA intermediate in protein synthesis containing a transcribed copy of the gene sequence that specifies the amino acid sequence of the polypeptide it encodes.

Metabolism

- The sum total of the chemical processes that take place in living organisms, resulting in growth, development, and all other forms of energy transformation.

Metabolite

- One particular chemical intermediate generated in metabolism.

Metabolites

- Metabolites are the chemicals that are produced by the cells in the body when they break down sugars, fats, and proteins to make energy.

metachronous cancer

- A new primary cancer that develops months or years after a first primary cancer, either in the same or a different organ.

MGPT

- Genetic tests using next-generation sequencing to simultaneously analyze multiple genes; also called multigene panel test or multigene test.

Micro-organism

- An organism that can be seen only under a microscope, usually, an ordinary light microscope. It includes bacteria, mycoplasm, yeasts, single-celled algae and protozoa. Multicellular organisms are normally not included, nor fungi apart from yeasts. Viruses are also not automatically included, as many scientists do not classify them as organisms.

Microdissection testicular sperm extraction (MicroTESE)

- A surgical procedure using an operating microscope to identify seminiferous tubules that may contain sperm to be extracted for IVF and/or ICSI.

Microepididymal sperm aspiration (MESA)

- Outpatient microsurgical procedure used to collect sperm in men with blockage of the male reproductive ducts such as prior vasectomy or absence of the vas deferens. Used in IVF-ICSI procedures.

Micromanipulation

- Technology that allows micro-operative procedures to be performed on the spermatozoon, oocyte, zygote, or embryo.
- The IVF laboratory process whereby the egg or embryo is held with special instruments and surgically altered by procedures such as intracytoplasmic sperm injection (ICSI), assisted hatching, or embryo biopsy.

Micromanipulation in ART

- A micro-operative ART procedure performed on sperm, egg or embryo; the most common ART micromanipulation procedures are ICSI, assisted hatching and gamete or embryo biopsy for PGT.

microsatellite

- Repetitive DNA segments scattered throughout the genome, often used as genetic markers due to their high variability in repeat number between individuals.

microsatellite instability

- A condition where errors in DNA mismatch repair lead to variations in microsatellite lengths; also called MSI.

Microsurgical epididymal sperm aspiration/ extraction (MESA/MESE)

- A surgical procedure performed with the assistance of an operating microscope to retrieve sperm from the epididymis of men with obstructive azoospermia. In the absence of optical magnification, any surgical procedure to retrieve sperm from the epididymis should also be registered as MESE.

MicroTESE

- Acronym for Microsurgical testicular sperm extraction.

Mild ovarian stimulation for IVF

- A procedure in which the ovaries are stimulated with gonadotropins and/or other compounds, with the intent to limit the number of oocytes obtained for IVF to fewer than seven.
- A protocol in which the ovaries are stimulated with gonadotropins, and/or other pharmacological compounds, with the intention of limiting the number of oocytes following stimulation for IVF.

Miscarriage (also called spontaneous abortion)

- A pregnancy ending in the spontaneous loss of the embryo or fetus before 20 weeks of gestation.

Missed abortion

- A clinical abortion where the embryo(s) or fetus(es) is/are nonviable and is/are not expelled spontaneously from the uterus.

Missed spontaneous abortion/missed miscarriage

- Spontaneous loss of a clinical pregnancy before 22 completed weeks of gestational age, in which the embryo(s) or fetus(es) is/are nonviable and is/are not spontaneously absorbed or expelled from the uterus.

missense mutation

- A genetic alteration where a single nucleotide substitution results in a different amino acid in a protein; also called missense variant.

missense variant

- A genetic change caused by a single nucleotide substitution that results in a different amino acid; also called missense mutation.

Mitochondria

- Membrane-bound cellular organelles in which organic substrates derived from food are oxidized to provide energy for all kinds of vital activities. They carry their own complement of DNA and are replicated independently so that when the cell divides, each daughter cell will receive half of the mitochondria.

Mitochondria/Mitochondrion

- Organelles in the cytoplasm of eukaryotic cells involved in energy production, typically shaped like kidney beans.

Mitochondrial DNA

- DNA located in the mitochondria, typically a circular chromosome. In humans, it has about 16,600 base pairs and encodes approximately 37 genes.

MLPA

- Multiplex ligation-dependent probe amplification, a laboratory method used to detect copy number changes in genomic sequences; also called MLPA.

Mobile Genetic Element, also called Transposon or Transposable Genetic Element

- A sequence of DNA that can move (transpose) from one place to another in the genome of a cell.

mode of inheritance

- The manner in which a genetic trait or disorder is transmitted from one generation to the next, such as autosomal dominant, autosomal recessive, or X-linked inheritance.

Modified natural cycle

- An ART procedure in which one or more oocytes are collected from the ovaries during a spontaneous menstrual cycle. Pharmacological compounds are administered with the sole purpose of blocking the spontaneous LH surge and/or inducing final oocyte maturation.
- An IVF procedure in which one or more oocytes are collected from the ovaries during a spontaneous menstrual cycle. Drugs are administered with the sole purpose of blocking the spontaneous LH surge and/or inducing final oocyte maturation.

Monosomy

- The absence of one of the two homologous chromosomes in embryos. Autosomal monosomies in embryos are not compatible with life. Embryos with sex chromosome monosomies are rarely compatible with life.

Morula

- An embryo formed after completion of compaction, typically 4 days after insemination or ICSI.

mosaicism

- The presence of two or more genetically distinct cell lines within an individual or tissue.

Mosaicism

- A state in which there is more than one karyotypically distinct cell population arising from a single embryo.
- The presence of more than one genetic cell line in an organism, all derived from the same zygote. If cells come from different zygotes, the organism is a chimera.
- When a biopsied embryo is found to contain cells from two cell lines where at least one is genetically abnormal.

Motile

- Moving.

MSAFP

- Maternal serum alpha-fetoprotein screening, a test measuring alpha-fetoprotein in a pregnant woman's blood to screen for Down syndrome or neural tube defects.

MSI

- Microsatellite instability; a marker of abnormal DNA mismatch repair leading to variable microsatellite lengths; also called MSI.

mucosal neuroma

- A benign tumor of nerve tissue found on the tongue, lips, or gastrointestinal tract, characteristic of multiple endocrine neoplasia type 2B (MEN2B).

Multifetal pregnancy reduction

- Also known as selective reduction. A procedure to reduce the number of fetuses in the uterus. This procedure is sometimes performed on women who are pregnant with multiple fetuses who are at an increased risk of late miscarriage or premature labor. These risks increase with the number of fetuses.

Multigene families

- Genes that exist in multiple copies in the genome, from several copies to many thousands or hundreds of thousands of copies.

multigene panel test

- Genetic tests that analyze multiple genes simultaneously using next-generation sequencing; also called MGPT or multigene test.

multigene test

- Genetic tests that analyze multiple genes simultaneously using next-generation sequencing; also called MGPT or multigene panel test.

Multinucleation

- The presence of more than one nucleus in a cell.

Multiple birth

- The complete expulsion or extraction from a woman of more than one fetus, after 22 completed weeks of gestational age, irrespective of whether it is a live birth or stillbirth. Births refer to the individual newborn; for example, a twin delivery represents two births.

Multiple gestation

- A pregnancy with more than one embryo or fetus.

Multiple gestation/birth

- A pregnancy/delivery with more than one fetus/neonate.

Multiple live-birth delivery

- The delivery of more than one infant with at least one born alive.

multiplex ligation-dependent probe amplification

- A laboratory technique to detect unusual copy number changes (insertions or deletions) in genomic sequences; also known as MLPA.

Mutagen

- A substance or agent that causes genetic mutations, or chemical alteration of the genetic material, DNA.

Mutation

- A permanent change in the DNA sequence in a cell or organism. Mutations can be harmful, beneficial, or neutral, and may occur in germ cells or somatically.

mutation

- A change in the usual DNA sequence at a specific gene locus. Mutations can be harmful, beneficial, or neutral. The term 'variant' is sometimes used synonymously.

mutation analysis

- A genetic testing method aimed at detecting a specific variant, panel of variants, or type of variant in a gene, distinct from complete gene sequencing.

nail dystrophy

- Distortion or malformation of the fingernails and toenails; also called onychodystrophy.

Nash equilibrium

- A stable position in a game. For example, in a simple coordination game, if everyone drives on the right side of the road, that is a Nash equilibrium.

Natural cycle ART

- An ART procedure in which one or more oocytes are collected from the ovaries during a menstrual cycle without the use of any pharmacological compound.

Natural cycle IVF

- An IVF procedure in which one or more oocytes are collected from the ovaries during a spontaneous menstrual cycle without any drug use.

Necrozoospermia

- The description of an ejaculate in which no live spermatozoa can be found.

negative predictive value

- The likelihood that an individual with a negative test result is truly unaffected and does not carry the specific gene mutation; also called NPV.

Neonatal death

- Death of a live born baby within 28 days of birth. Neonatal period: the time interval that commences at birth and ends 28 completed days after birth.

Neonatal death/mortality

- Death of a live born baby within 28 days of birth. This can be sub-divided into a) early, if death occurs in the first 7 days after birth; and b) late, if death occurs between 8 and 28 days after birth.

Neonatal mortality rate

- Number of neonatal deaths (up to 28 days) per 1000 live births.

Neonatal period

- The period which commences at birth and ends at 28 completed days after birth.

network effect

- The effect that an additional user of a product has on others who use it. Network effects can be positive or negative; for example, positive feedback loops occur when more people using a product make others better off.

new mutation

- A genetic alteration present for the first time in a family member due to a variant in a parent's germ cell or in the fertilized egg; also called de novo mutation, de novo variant, or new variant.

new variant

- A genetic alteration present for the first time in a family member due to a variant in a parent's germ cell or in the fertilized egg; also called de novo mutation, de novo variant, or new mutation.

next-generation sequencing

- A high-throughput method for sequencing DNA that processes multiple sequences in parallel; also called massively parallel sequencing and NGS.

NGS

- Next-generation sequencing; a high-throughput DNA sequencing method that processes many sequences in parallel; also known as massively parallel sequencing.

NIPT

- Noninvasive prenatal testing, a technique for assessing fetal DNA from maternal blood without invasive procedures.

Non-obstructive azoospermia

- Absence of spermatozoa in the ejaculate due to lack of production of mature spermatozoa.

noncarrier

- An individual who does not carry a mutation that has been identified in their family.

noninferiority study

- A research study designed to determine whether a new intervention is not worse than a control by a predetermined margin.

nonpenetrance

- The phenomenon in which an individual carries a pathogenic genetic variant but does not express the associated phenotype.

nonsense mutation

- A genetic alteration that creates a premature stop codon, leading to a truncated, usually nonfunctional protein; also called nonsense variant.

nonsense variant

- A genetic change that produces a premature stop codon, resulting in a truncated, usually nonfunctional protein; also called nonsense mutation.

novel mutation

- A newly discovered, distinct genetic alteration that is not the same as a new or de novo mutation; also called novel variant.

novel variant

- A newly discovered, distinct genetic alteration that is not the same as a new or de novo mutation; also called novel mutation.

NPV

- Negative predictive value; the likelihood that an individual with a negative test result is truly unaffected; also called negative predictive value.

Nuclear DNA

- The DNA contained within a cell's nucleus, as opposed to mitochondrial DNA.

Nuclear maturation

- The process during which the oocyte resumes meiosis and progresses from prophase I to metaphase II.

Nuclear transplant cloning

- A process whereby the nucleus containing the genome of an adult cell is transferred into an egg from which the nucleus was previously removed. The egg with the transplant nucleus is then stimulated to divide and develop into an organism.

nucleotide

- A molecule that is the basic building block of DNA and RNA, composed of a nitrogenous base, a phosphate group, and a sugar.

Nucleus

- A structure in the eukaryote cell bounded by a membrane, that contains the genetic material, in the form of DNA organized into chromosomes.
- The part of a eukaryotic cell enclosed by a nuclear membrane that contains the chromosomes.

null allele

- A variant of a gene that results in no functional gene product.

Obstructive azoospermia

- Absence of spermatozoa in the ejaculate due to occlusion of the ductal system.

Oligonucleotide

- A short stretch of DNA or RNA, typically twenty-five or fewer bases long, used in various DNA analysis techniques.

Oligospermia

- A term for low semen volume now replaced by hypospermia to avoid confusion with oligozoospermia.

Oligozoospermia

- Low concentration of spermatozoa in the ejaculate below the lower reference limit. When reporting results, the reference criteria should be specified.

Oncogenes

- Genes associated with cancer.

onychodystrophy

- Distortion or malformation of the fingernails and toenails; also called nail dystrophy.

Oocyte

- Medical term for egg, the female gamete. Also called ovum (singular) or ova (plural).
- The female gamete (egg).
- The female reproductive cell, also called an egg or ovum.

Oocyte aspiration

- Ovarian follicular aspiration performed with the aim of retrieving oocytes.

Oocyte bank

- Repository of cryopreserved oocytes stored for future use.

Oocyte cryopreservation

- The freezing or vitrification of oocytes for future use.

Oocyte donation

- The use of oocytes from an egg donor for reproductive purposes or research.

Oocyte donation cycle

- A cycle in which oocytes are collected from a donor for clinical application or research.
- An ART cycle in which oocytes are collected from an egg donor for reproductive purposes or research.

Oocyte maturation triggering

- An intervention intended to induce an oocyte in vitro or in vivo to resume meiosis to reach maturity (i.e. to reach metaphase II).

Oocyte recipient cycle

- An ART cycle in which a woman receives oocytes from a donor, or her partner if in a same sex relationship, to be used for reproductive purposes.
- An ART cycle in which a woman receives oocytes from a donor.

Oolemma

- The cytoplasmic membrane enclosing the oocyte.

Ooplasm

- The cytoplasm of the oocyte.

Operon

- A multigenic unit consisting of a regulator gene, a promoter-operator region and one or more other genes whose expression is regulated.

oral papilloma

- A benign epithelial growth in the mouth or on the lips, often associated with Cowden syndrome.

ORF

- An open reading frame, the part of a DNA sequence that can be transcribed into mRNA and potentially translated into protein, starting with a start codon and ending with a stop codon.

Organelle

- A small, functional structure within a cell that is separated by its own membrane. Only eukaryotic cells have organelles.

Other reason, infertility

- Reason for using ART that is related to infertility; may include immunological problems, chromosomal abnormalities, cancer chemotherapy, and serious illnesses.

Other reason, non-infertility

- Reason for using ART that is not related to infertility.

Ovarian hyperstimulation syndrome

- (OHSS) An exaggerated systemic response to ovarian stimulation characterized by a wide spectrum of clinical and laboratory manifestations. It is classified as mild, moderate, or severe according to the degree of abdominal distention, ovarian enlargement, and respiratory, hemodynamic, and metabolic complications.
- A complication of ovarian stimulation that can cause enlarged ovaries, a distended abdomen, nausea, vomiting or diarrhea, fluid in the abdominal cavity or chest, breathing difficulties, changes in blood volume or viscosity, and diminished kidney perfusion and function.

Ovarian hyperstimulation syndrome (OHSS)

- A condition that may result from ovulation induction characterized by enlargement of the ovaries, fluid retention, and weight gain.
- An exaggerated systemic response to ovarian stimulation characterized by a wide spectrum of clinical and laboratory manifestations. It may be classified as mild, moderate or severe according to the degree of abdominal distention, ovarian enlargement and respiratory, hemodynamic and metabolic complications.

Ovarian reserve

- A term generally used to indicate the number and/or quality of oocytes, reflecting the ability to reproduce. Ovarian reserve can be assessed by any of several means. They include: female age; number of antral follicles on ultrasound; anti-Mullerian hormone levels; follicle stimulating hormone and estradiol levels; clomiphene citrate challenge test; response to gonadotropin stimulation, and oocyte and/or embryo assessment during an ART procedure, based on number, morphology or genetic assessment of the oocytes and/or embryos.
- A woman's fertility potential in the absence of specific pathophysiologic changes in her reproductive system. Diminished ovarian reserve is associated with depletion in the number of eggs and worsening of oocyte quality.

Ovarian stimulation

- See Ovulation induction.
- The use of drugs (oral or injected) to stimulate the ovaries to develop follicles and eggs.

Ovarian stimulation (OS)

- Pharmacological treatment with the intention of inducing the development of ovarian follicles. It can be used for two purposes: 1) for timed intercourse or insemination; 2) in ART, to obtain multiple oocytes at follicular aspiration.

Ovarian tissue cryopreservation

- The process of slow-freezing or vitrification of tissue surgically excised from the ovary with the intention of preserving reproductive capacity.

Ovarian torsion

- Partial or complete rotation of the ovarian vascular pedicle that causes obstruction to ovarian blood flow, potentially leading to necrosis of ovarian tissue.

Ovary (Ovaries)

- The two female sex glands in the pelvis, located one on each side of the uterus. The ovaries produce eggs and hormones including estrogen, progesterone, and androgens.

Ovulation

- Release of an egg from the ovary.
- The natural process of expulsion of a mature egg from its ovarian follicle.

Ovulation induction

- (OI) Pharmacologic treatment of women with anovulation or oligo-ovulation with the intention of inducing normal ovulatory cycles.
- The administration of hormone medications (ovulation drugs) that stimulate the ovaries to produce multiple eggs. Sometimes called enhanced follicular recruitment or controlled ovarian hyperstimulation.

Ovulation induction (OI)

- Pharmacological treatment of women with anovulation or oligo-ovulation with the intention of inducing normal ovulatory cycles.

Ovulatory dysfunction

- A diagnostic category used when a woman's ovaries are not producing eggs normally. It is usually characterized by irregular menstrual cycles with ovaries that are not producing one mature egg each month. It includes polycystic ovary syndrome (PCOS) and multiple ovarian cysts.

palmar pit

- A small depression in the skin on the palm of the hand. Depressions may also be seen on the sole of the foot (plantar pits). They are usually pink or flesh-colored and range from 2 to 3 mm in diameter and 1 to 3 mm in depth. Palmar pits are often more visible after a brief soaking of the hands or feet. This condition is associated with basal cell nevus syndrome, which is a hereditary syndrome associated with the development of basal cell carcinoma.

Parthenogenetic activation

- The process by which an oocyte is activated to undergo development in the absence of fertilization.

Parthenote

- The product of an oocyte that has undergone activation in the absence of the paternal genome, with (induced) or without (spontaneous) a purposeful intervention.

Pathogen

- Any agent that can cause disease.

Pathogenic

- Causing disease; in genetics, it refers to a variant that can cause disease, though not all pathogenic variants always manifest due to factors like penetrance.

pathogenic variant

- A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or disorder. When such a variant (or mutation) is inherited, development of symptoms is more likely, but not certain. Also called deleterious mutation, disease-causing mutation, predisposing mutation, and susceptibility gene mutation.

pathognomonic

- Findings that are distinctive or characteristic of a particular disease or condition and can be used to make a diagnosis.

pathological altruism

- Situations in which the attempt to promote others' welfare produces harms or inadvertently decreases the welfare of others.

Patient (nondonor) cycle

- An ART cycle where an embryo is formed from the egg of the patient and either partner or donor sperm and then transferred back to the patient.

PCR

- A common laboratory technique used during molecular genetic testing to produce many copies of a specific sequence of DNA. PCR allows these DNA sequences to be amplified so there is a sufficient quantity of DNA to be analyzed by molecular genetic tests. Also called polymerase chain reaction.

pedigree

- A diagram of family history that uses standardized symbols. A pedigree shows relationships between family members and indicates which individuals have certain genetic pathogenic variants, traits, and diseases within a family as well as vital status. It can be used to determine disease inheritance patterns within a family.

penetrance

- Penetrance refers to the likelihood that a clinical condition will occur when a particular genotype is present. For example, the penetrance for breast cancer in female carriers of BRCA1 pathogenic variants is often quoted by age 50 years and by age 70 years.

Penetrance

- The percentage of the time a particular DNA variant results in the associated disease or trait.

Penile vibratory stimulation (PVS)

- A procedure to cause ejaculation of sperm, performed by vibratory stimulation of the penis.

Percutaneous epididymal sperm aspiration (PESA)

- A sperm aspiration procedure in which a needle is inserted into the epididymis (gland that carries sperm from testicle to vas deferens) in order to retrieve sperm for use in an IVF procedure.
- A surgical procedure in which a needle is introduced percutaneously into the epididymis with the intention of obtaining sperm.

Perinatal death/mortality

- Fetal or neonatal death occurring during late pregnancy (at 22 completed weeks of gestational age and later), during childbirth, or up to seven completed days after birth.

Perinatal mortality

- Fetal or neonatal death occurring during late pregnancy (at 20 completed weeks of Gestational age GA and later), during childbirth, or up to 7 completed days after birth.

Perinatal mortality rate

- The number of perinatal deaths per 1000 total births (stillbirths plus live births).

Period total fertility rate (PTFR)

- The estimated average number of live born children per woman that would be born to a cohort of women throughout their reproductive years, if the fertility rates by age in a given period remained constant at the current age-specific fertility rate.

Perivitelline space

- The space between the cytoplasmic membrane enclosing the oocyte and the innermost layer of the zona pellucida. (This space may contain the first and second polar bodies and extracellular fragments.)

PESA

- An acronym for percutaneous epididymal sperm aspiration.

PGD

- Preimplantation genetic diagnosis, the process of genetically testing one or more cells from an embryo to determine its likely traits before transfer.

PGT (preimplantation genetic testing)

- Diagnostic or screening techniques performed on embryos before transfer to (1) detect specific genetic conditions to reduce the risk of passing inherited diseases to children or (2) screen for an abnormal number of chromosomes, which is more frequent for patients with advanced age, recurrent miscarriages, or prior failed IVF.

PGT-A

- Testing for Aneuploidy, the presence of too many or too few chromosomes in an embryo.

PGT-M

- Testing for Monogenic disorders which are single gene changes carried by the biologic mother and/or father, e.g. cystic fibrosis, sickle cell disease).

PGT-SR

- Testing for Structural Rearrangements of chromosomes which are chromosomal variations that are carried by the biologic mother and/or father and increase the risk of miscarriage. These rearrangements or translocations are often found in patients with recurrent miscarriage.

Phage

- A bacterial virus.

phenocopy

- A phenotypic trait or disease that resembles the trait expressed by a particular genotype but in an individual who is not a carrier of that genotype.

phenotype

- The observable characteristics or traits in an individual based on the expression of their genes. The phenotype is determined by the individual's genotype and may be influenced by environmental or other genetic factors.

Phenotype

- An individual's observable traits, including physical characteristics and biochemical properties, resulting from the interaction of genotype and environment.
- Phenotype is how a person looks (on the outside and inside the body) due to his or her genes and the environment (for example, having a certain eye color, being a specific blood type, or being a certain height). Phenotype also can refer to how a person's body functions, for example, whether he or she has a certain disease.
- The expressed characteristics, or an expressed character of an organism due to its genotype.

Pituitary down-regulation

- A medical or pharmacological method to prevent the release of gonadotropins (FSH, LH) from the pituitary gland.

Pituitary gland

- A small gland just beneath the hypothalamus in the brain that secretes follicle stimulating hormone (FSH) and luteinizing hormone (LH).

plantar pit

- A small depression in the skin on the sole of the foot. Depressions may also be seen on the palm of the hand (palmar pits). They are usually pink or flesh-colored and range from 2 to 3 mm in diameter and 1 to 3 mm in depth. Plantar pits are often more visible after a brief soaking. This condition is associated with basal cell nevus syndrome, which is a hereditary syndrome associated with the development of basal cell carcinoma.

Plasmid

- A piece of parasitic genetic material found in a cell that can propagate itself using the cell's energetic resources.

pluripotent stem cell

- A cell capable of becoming any kind of specific cell, such as a blood, skin, bone, or sperm cell. Induced pluripotent stem cells are derived by taking an adult cell and turning it into a pluripotent stem cell.

poikiloderma

- Irregular patterns of reddish brown pigmentation of the skin associated with sun exposure, aging, or a genetic cause.

point mutation

- A genetic alteration caused by the substitution of a single nucleotide for another nucleotide. Also called point variant.

point variant

- A genetic alteration caused by the substitution of a single nucleotide for another nucleotide. Also called point mutation.

Polar bodies

- The small bodies containing chromosomes segregated from the oocyte by asymmetric division during telophase. The first polar body is extruded at telophase I and normally contains only chromosomes with duplicated chromatids (2c); the second polar body is extruded in response to fertilization or in response to parthenogenetic activation and normally contains chromosomes comprising single chromatids (1c).

Polycystic ovary (PCO)

- An ovary with at least 12 follicles measuring 2–9 mm in diameter in at least one ovary (Rotterdam criteria). PCO may be present in women with PCOS, but also in women with normal ovulatory function and normal fertility.

Polycystic ovary syndrome (PCOS)

- A heterogeneous condition, which requires the presence of two of the following three criteria: (1) Oligo-ovulation or anovulation; (2) Hyperandrogenism (clinical evidence of hirsutism, acne, alopecia and/or biochemical hyperandrogenemia); (3) Polycystic ovaries, as assessed by ultrasound scan with more than 24 total antral follicles (2–9 mm in size) in both ovaries.

Polygenes

- The (hypothetical) many genes affecting a character, each having a small, additive effect on the character.

polygenic risk score

- A way of gauging the likelihood that an embryo will develop a trait, given that the trait involves many different genes interacting with one another.
- An assessment of the risk of a specific condition based on the collective influence of many genetic variants. Also called PRS.

polymerase chain reaction

- A common laboratory technique used during molecular genetic testing to produce many copies of a specific sequence of DNA. Also called PCR.

polymorphism

- A common variant in a specific sequence of DNA, typically defined as an allele frequency of at least 1% in the general population.

Polymorphism

- A common genomic variation in a population, typically defined as having an allele frequency of at least 1%.

Polypeptide or Protein

- A long chain of different amino-acids joined together by special chemical (peptide) bonds.

Polyploidy

- The condition in which a cell has more than two haploid sets of chromosomes: e.g. a triploid embryo has three sets of chromosomes and a tetraploid embryo has four sets. Polyploidy in a human embryo is not compatible with life.

Polyps

- A general term that describes any mass of tissue that bulges or projects out or upward from the normal surface level.

Polyspermy

- The process by which an oocyte is penetrated by more than one spermatozoon.

Poor ovarian responder (POR) in assisted reproductive technology

- A woman treated with ovarian stimulation for ART, in which at least two of the following features are present: (1) Advanced maternal age (40 years); (2) A previous poor ovarian response (3 oocytes with a conventional stimulation protocol aimed at obtaining more than three oocytes); and, (3) An abnormal ovarian reserve test (i.e. antral follicle count 5–7 follicles or anti-Mullerian hormone 0.5–1.1 ng/ml (Bologna criteria); or other reference values obtained from a standardized reference population.)

Poor ovarian response (POR) to ovarian stimulation

- A condition in which fewer than four follicles and/or oocytes are developed/obtained following ovarian stimulation with the intention of obtaining more follicles and oocytes.

positive predictive value

- The likelihood that an individual with a positive test result truly has the particular gene and/or disease in question. Also called PPV.

Post-implantation embryo

- An embryo at a stage of development beyond attachment to the endometrium to eight completed weeks after fertilization, which is equivalent to 10 weeks of gestational age.

Post-term birth

- A live birth or stillbirth that takes place after 42 completed weeks of gestational age.

Posthumous reproduction

- A process utilizing gametes and/or embryos from a deceased person or persons with the intention of producing offspring.

PPV

- The likelihood that an individual with a positive test result truly has the particular gene and/or disease in question. Also called positive predictive value.

Pre-implantation embryo

- An embryo at a stage of development beginning with division of the zygote into two cells and ending just prior to implantation into a uterus.

pre-implantation genetic testing (PGT)

- A procedure that allows us to scan the genome of an embryo before it is implanted in order to test its likelihood of developing specific traits. (The technical term for a pre-implantation embryo that hasn't yet divided is 'zygote', but 'embryo' is used informally to cover zygotes as well.)

predisposing mutation

- A genetic alteration that increases an individual's susceptibility or predisposition to a certain disease or disorder. Also called deleterious mutation, disease-causing mutation, pathogenic variant, and susceptibility gene mutation.

Pregnancy

- A state of reproduction beginning with implantation of an embryo in a woman and ending with the complete expulsion and/or extraction of all products of implantation.

Pregnancy loss

- The outcome of any pregnancy that does not result in at least one live birth. When reporting pregnancy loss, the estimated gestational age at the end of pregnancy should be recorded.

Pregnancy of unknown location (PUL)

- A pregnancy documented by a positive human chorionic gonadotropin (hCG) test without visualization of pregnancy by ultrasound. This condition exists only after circulating hCG concentration is compatible with ultrasound visualization of a gestational sac.

Preimplantation genetic diagnosis

- (PGD) Analysis of polar bodies, blastomeres, or trophectoderm from oocytes, zygotes, or embryos for the detection of specific genetic, structural, and/or chromosomal alterations.

Preimplantation genetic diagnosis (PGD) and screening (PGS)

- These terms have now been replaced by preimplantation genetic testing PGT. (See term PGT and its definitions.)

Preimplantation genetic screening

- (PGS) Analysis of polar bodies, blastomeres, or trophectoderm from oocytes, zygotes, or embryos for the detection of aneuploidy, mutation, and/or DNA rearrangement.

Preimplantation genetic testing (PGT)

- A test performed in which cells are removed from an embryo. The cells are then screened for genetic abnormalities. May be performed in conjunction with IVF. There are several types of testing that can be performed.
- A test performed to analyze the DNA from oocytes (polar bodies) or embryos (cleavage stage or blastocyst) for HLA-typing or for determining genetic abnormalities. These include: PGT for aneuploidies (PGT-A); PGT for monogenic/single gene defects (PGT-M); and PGT for chromosomal structural rearrangements (PGT-SR).

Premature ejaculation

- A condition in which semen is released sooner than desired.

Premature ovarian insufficiency

- A condition characterized by hypergonadotropic hypogonadism in women younger than age 40 years (also known as premature or primary ovarian failure). It includes women with premature menopause.

presymptomatic testing

- Genetic testing performed in an individual who does not show symptoms of the disorder but is at risk of developing it.

Preterm birth

- A birth that takes place after 22 weeks and before 37 completed weeks of gestational age.
- A live birth or stillbirth that takes place after at least 20 but before 37 completed weeks of gestational age.

Primary childlessness

- A condition in which a person has never delivered a live child, or has never been a legal or societally-recognized parent to a child.

Primary female infertility

- A woman who has never been diagnosed with a clinical pregnancy and meets the criteria of being classified as having infertility.

Primary involuntary childlessness

- A condition in a person with a child wish, who has never delivered a live child, or has never been a legal or societally-recognized parent to a child. A major cause of primary involuntary childlessness is infertility.

Primary male infertility

- A man who has never initiated a clinical pregnancy and meets the criteria of being classified as infertile.

Pro-nuclear transfer

- (PNT) The pro-nucleus is the nucleus of a sperm or an egg cell during the process of fertilisation after the sperm enters the egg, but before they fuse. PNT involves removing the pro-nuclei (nuclear material) from a newly fertilized egg that has unhealthy mitochondria. The pro-nuclei are then transferred into a donated embryo, with healthy mitochondria, that has had its own, original pro-nuclei removed.

proband

- The first individual in a family to be identified as possibly having a genetic disorder or condition.

procreative altruism

- The moral principle that parents should try to create children whose existence can be expected to contribute more to the well-being of others than any alternative child they could have.

procreative beneficence

- The moral principle that parents should try to create children with the best chance of the best life, given the available information.

Progesterone

- A female hormone secreted during the second half of the menstrual cycle. It prepares the lining of the uterus for implantation of a fertilized egg.

Prokaryote

- The class of living things, including all bacteria, that do not have a nucleus in their cell.

Promoter

- A piece of genetic material that acts as a gene switch, so that a gene can become expressed in the cell. It is the region at which the RNA polymerase binds to start transcription. Most promoters are located upstream of the gene, except that some eukaryotic genes have promoters internal to the gene.

Pronuclei

- The nuclei of the male and female gametes (sperm and egg) seen in the one-cell embryo (zygote).

Pronuclei transfer

- Transfer of the pronuclei from a patient's zygote to an enucleated donated zygote.

Pronucleus

- A round structure in the oocyte surrounded by a membrane containing chromatin. Normally, two pronuclei are seen after fertilization, each containing a haploid set of chromosomes, one set from the oocyte and one from the sperm, before zygote formation.

Protein

- A protein is made up of building blocks called amino acids. The main role of DNA is to act as the instructions for making proteins. It is actually proteins that make up most of the structures in our bodies and perform most of life's functions. For example, proteins make up hair and skin. Proteins in our eyes change shape in response to light so we can see. Proteins in our bodies break down food. Proteins are made in cells and are the major parts of cells, which are the vital working units of all living things.

Proto-oncogenes

- Cellular genes which, when mutated, or over-expressed become oncogenes.

Provirus

- A virus that has inserted its genome or a complementary copy of its genome into the host cell genome.

PRS

- An assessment of the risk of a specific condition based on the collective influence of many genetic variants. Also called polygenic risk score.

Pseudogene

- A nonfunctional stretch of DNA that resembles a functional gene, often related by descent.

pseudogene

- A DNA sequence that resembles a gene but has been mutated into an inactive form over the course of evolution. Pseudogenes do not produce functional proteins, although some may have regulatory effects.

public good

- A good or outcome that is freely available to all once produced, as opposed to private goods that allow owners to exclude others.

Recessive

- A Mendelian trait that is expressed only when an individual has two copies of an allele.
- For recessive diseases, both copies of a gene must have the DNA mutation for a person to have one of these diseases. If both parents have one copy of the mutated gene, each child has a 25% chance of having the disease, even though neither parent has it. In such cases, each parent is called a carrier of the disease. They can pass the disease on to their children, but do not have the disease themselves.

Recessive allele

- An allele which is not expressed unless two copies are present in the individual, i.e., in homozygous condition.

Recipient (ART)

- A person or couple who receives donated eggs, sperm or embryos for the purposes of initiating a pregnancy with the intention of becoming a legally recognized parent.

Recipient ART cycle

- An ART cycle in which a woman receives zygote(s) or embryo(s) from donor(s) or a partner.

Recombination

- The formation of new combinations of alleles or new genes which occur when two homologous DNA or chromosomes break and exchange parts.

Recombination hotspot

- A frequent breakpoint in the DNA or RNA, which promotes recombination.

recurrence risk

- The likelihood that a hereditary trait or disorder present in one family member will occur again in other family members.

Recurrent spontaneous abortion/miscarriage

- The spontaneous loss of two or more clinical pregnancies prior to 22 completed weeks of gestational age.
- The spontaneous loss of two or more clinical pregnancies.

reduced penetrance

- A situation where some individuals who carry a pathogenic variant do not express the associated trait; also called incomplete penetrance.

Reductionism

- The doctrine that complex systems can be completely understood in terms of its simplest parts. For example, an organism is to be completely understood in terms of its genes, a society in terms of its individuals, and so on.

REI

- Reproductive endocrinology and infertility, a medical subspecialty focusing on assisted reproduction methods like IVF.

Reproductive surgery

- Surgical procedures performed to diagnose, conserve, correct and/or improve reproductive function in either men or women. Surgery for contraceptive purposes, such as tubal ligation and vasectomy, are also included within this term.
- Surgical procedures performed to diagnose, conserve, correct, and/or improve reproductive function.

Retrograde ejaculation

- A condition that causes the semen to be forced backward from the ejaculatory ducts into the bladder during ejaculation.

Retrotransposon

- A mobile genetic element that depends on a reverse transcription step to move and to duplicate.
- A transposable element that moves by first being transcribed into RNA and then reverse-transcribed back into DNA at a new location.

Reverse Transcription

- The reverse of transcription - making a copy of complementary DNA (cDNA) from an RNA sequence - catalyzed by the enzyme, reverse transcriptase.

Ribosomal RNA

- RNA molecules which make up the ribosome.

Ribosome

- An organelle in the cell required for protein synthesis.

risk assessment

- The quantitative or qualitative evaluation of an individual's risk of carrying a certain gene mutation, developing a particular disorder, or having a child with a certain disorder.

RNA

- Ribonucleic acid, a nucleic acid similar to DNA but using ribose and uracil, involved in gene expression and protein synthesis.
- RiboNucleic Acid, similar to DNA except for the sugar in the nucleotide unit, which is ribose, instead of deoxyribose, and the base Uracil instead of Thymine. RNA is the genetic material for RNA viruses.

RNA editing

- The process in which the base sequence of the RNA transcript is changed by addition of bases to the RNA molecule or by chemical transformation of one base to another. This subverts the genetic information carried in the genes.

RNA polymerase

- An enzyme that makes RNA.

RNA splicing

- The process by which introns are removed from the primary mRNA transcript and exons are joined together to form mature mRNA.

Salpingectomy

- The surgical removal of an entire Fallopian tube.

Salpingitis isthmica nodosa (SIN)

- A nodular thickening of the proximal Fallopian tube (where the tubes join the uterus), which can distort or occlude the tubes and increase the risk of ectopic pregnancy and infertility.

Salpingostomy

- A surgical procedure in which an opening is made in the Fallopian tube either to remove an ectopic pregnancy or open a blocked fluid-filled tube (hydrosalpinx).

Sanger sequencing

- A low-throughput laboratory method used to determine the nucleotide sequence of a piece of DNA by amplifying the target region with PCR and then sequencing the PCR products.

SCNT

- Somatic cell nuclear transfer, a cloning method where the nucleus of one cell is transferred into an enucleated egg, as in the cloning of Dolly the sheep.

screening

- The process of checking for disease in individuals who have no symptoms, which may include genetic testing to assess risk of developing an inherited disease.

SDR

- A second-degree relative: an aunt, uncle, grandparent, grandchild, niece, nephew, or half-sibling.

sebaceoma

- A benign yellow or flesh-colored lesion originating from an oil gland in the skin, typically found on the face or neck. May be associated with Muir-Torre syndrome. Also called sebaceous epithelioma.

sebaceous epithelioma

- A benign yellow or flesh-colored lesion originating from an oil gland in the skin, typically found on the face or neck. May be associated with Muir-Torre syndrome. Also called sebaceoma.

second-degree relative

- An aunt, uncle, grandparent, grandchild, niece, nephew, or half-sibling. Also called SDR.

Secondary female infertility

- A woman unable to establish a clinical pregnancy but who has previously been diagnosed with a clinical pregnancy.

Secondary involuntary childlessness

- A condition in a person with a child wish, who has previously delivered a live child, or is or has been a legal or societally-recognized parent to a child. A major cause of secondary involuntary childlessness is infertility.

Secondary male infertility

- A man who is unable to initiate a clinical pregnancy, but who had previously initiated a clinical pregnancy.

segregation analysis

- The process of fitting formal genetic models to family data to determine the most likely mode of inheritance for a trait or disease.

Semen

- The fluid ejaculated by the male.

Semen analysis

- A description of the ejaculate to assess function of the male reproductive tract. Characteristic parameters include volume, pH, concentration, motility, vitality, morphology of spermatozoa and presence of other cells.

Semen liquefaction

- The process whereby proteolytic enzymes degrade proteins causing seminal plasma to liquefy.

Semen viscosity

- The description of the relative fluidity of seminal plasma.

Semen volume

- The amount of fluid in an ejaculate.

Semen/Ejaculate

- The fluid at ejaculation that contains the cells and secretions originating from the testes and sex accessory glands.

Seminal plasma

- The fluids of the ejaculate.

sensitivity

- The frequency with which a test correctly identifies individuals who have the disease or genetic variant (true positive rate).

Septum, uterine

- A band of fibrous tissue present from birth that forms a wall within the uterine cavity. A septum may increase the risk of miscarriage and other pregnancy complications.

Sertoli cell

- The non-germinal cell type in the seminiferous tubule that mediates the actions of testosterone and FSH in the testis, provides nutrients and proteins to the developing spermatogenic cells, creates the blood-testis barrier, and secretes Mullerian-inhibiting hormone.

Sertoli cell-only syndrome

- A condition in which only Sertoli cells line the seminiferous tubules with usually a complete absence of germ cells; also referred to as germ cell aplasia. Spermatogenesis in isolated foci can be observed in rare cases.

SET (single embryo transfer)

- A procedure where one embryo, regardless of how many embryos are available, is transferred. The embryo selected for SET might be a frozen (cryopreserved) embryo from a previous IVF cycle or a fresh embryo from the current fresh IVF cycle.

Severe ovarian hyperstimulation syndrome

- Severe OHSS is defined to occur when hospitalization is indicated. (See definition of ovarian hyperstimulation syndrome.)

Severe ovarian hyperstimulation syndrome (OHSS)

- A systemic response as a result of ovarian stimulation interventions that is characterized by severe abdominal discomfort and/or other symptoms of ascites, hemoconcentration (Hct > 45) and/or other serious biochemical abnormalities requiring hospitalization for observation and/or for medical intervention (paracentesis, other).

Sex chromosomes

- Chromosomes that determine an organism's sex; in mammals, typically the X and Y chromosomes.

SINES

- Short interspersed elements, short DNA sequences inserted into the genome by transposons or retrotransposons, often considered 'junk' DNA.

Single embryo transfer (SET)

- The transfer of one embryo in an ART procedure. Defined as elective (eSET) when more than one embryo of sufficient quality for transfer is available.

single nucleotide polymorphism

- A DNA sequence variation caused by a single nucleotide difference from the reference sequence. Also called SNP.

Single Nucleotide Polymorphism (SNP)

- Single nucleotide polymorphisms (SNPs) are changes at a single DNA base that are present among at least 1% of people in at least one population. For example, at a given DNA location, some people will have one base (e.g., adenine), while other people will have a different base (e.g., guanine). The SNP that is more common among a given group of people is called the major allele and the one that is less common is called the minor allele.

single nucleotide variant

- A DNA sequence variation caused by a single nucleotide change. Also called SNV.

Singleton live-birth delivery

- The delivery of a single infant born alive. Singleton live-birth delivery does not include the delivery of more than one infant with only one born alive.

sister chromatid exchange

- The exchange of genetic material between sister chromatids during mitosis.

skin tag

- A small, benign skin growth that may have a stalk (peduncle). Also called acrochordon.

Slow-freezing

- A cryopreservation procedure in which the temperature of the cell(s) is lowered in a stepwise fashion, typically using a computer controlled rate, from physiological (or room) temperature to extreme low temperature.

Small for gestational age

- A birth weight less than the 10th centile for gestational age. When reporting results the reference criteria should be specified. If gestational age is unknown, the birth weight should be registered.
- Birth weight less than 2 standard deviations below the mean or less than the 10th percentile according to local intrauterine growth charts.

SNP

- A single nucleotide polymorphism; a DNA sequence variation that occurs when one nucleotide differs from the reference sequence.
- Single nucleotide polymorphism, a common variation in DNA at a single base pair.

SNP chip

- An array containing hundreds of thousands or millions of SNPs, allowing fast, inexpensive analysis of a DNA sample for its SNPs.

SNV

- A single nucleotide variant; a DNA sequence variation that occurs when a single nucleotide in the genome is altered.

Society for Assisted Reproductive Technology (SART)

- A society affiliated with the ASRM and comprised of representatives from assisted reproductive technology programs who have demonstrated their ability to perform IVF.

Somaclonal variation

- Genetic variations of plant cells arising in cell culture, due to enhanced genetic instability.

somatic mutation

- A mutation that occurs in somatic cells (non-germline) and is not inherited by offspring. Also called somatic variant.

somatic variant

- A DNA variant occurring in somatic cells that is not present in the germline; also called somatic mutation.

Southern blot

- An electrophoresis-based technique used to detect large deletions in DNA that may be missed by PCR-based methods.

specificity

- The frequency with which a test correctly identifies individuals who do not have the disease or genetic variant (true negative rate).

Sperm

- The male reproductive cell.
- The male reproductive cells that fertilize a woman's egg. The sperm head carries genetic material (chromosomes), the midpiece produces energy for movement, and the long, thin tail wiggles to propel the sperm.

Sperm bank

- Repository of cryopreserved sperm stored for future use.

Sperm concentration

- The (measure of the) number of spermatozoa in millions per 1 ml of semen.

Sperm density

- A measure of the mass/volume ratio (specific gravity) for spermatozoa.

Sperm isolation

- A procedure that involves the separation of sperm through centrifugation and resuspension in culture media. It can be used to remove seminal plasma and infectious agents before IUI and ART procedures. This procedure has been shown to be effective in the removal of HIV. It may also be effective in removing other infectious particles but clinical safety and efficacy have to be established for each particular infection. This term is sometimes referred to as 'sperm washing'.

Sperm motility

- The percentage of moving spermatozoa relative to the total number of spermatozoa.

Sperm recipient cycle

- A MAR cycle in which a woman receives spermatozoa from a person who is not her sexually intimate partner. In the case of ART registry data, a sperm recipient cycle would only include data from cycles using ART procedures.
- An ART cycle in which a woman receives spermatozoa from a donor who is not her partner.

Sperm vitality

- The percentage of live spermatozoa relative to the total number of spermatozoa.

Spermatogenic arrest

- Failure of germ cells to progress through specific stages of spermatogenesis at onset or during meiosis.

Spermatozoon

- The mature male reproductive cell produced in the testis that has the capacity to fertilize an oocyte. A head carries genetic material, a midpiece produces energy for movement, and a long, thin tail propels the sperm.

Spina bifida

- A birth defect of the spinal column. Spina bifida is the failure of the spine to close properly during development.

splice-site mutation

- A mutation at the junction of an exon and an intron that disrupts RNA splicing, potentially causing loss of exons or inclusion of introns; also called splice-site variant.

splice-site variant

- A mutation at the junction of an exon and an intron that disrupts RNA splicing, potentially causing loss of exons or inclusion of introns; also called splice-site mutation.

Spontaneous abortion

- See Miscarriage.

Spontaneous abortion/miscarriage

- The spontaneous loss of a clinical pregnancy before 20 completed weeks of gestational age (18 weeks after fertilization) or, if gestational age is unknown, the loss of an embryo/fetus of less than 400 g.
- The spontaneous loss of an intra-uterine pregnancy prior to 22 completed weeks of gestational age.

Spontaneous reduction/vanishing sac(s)

- The spontaneous disappearance of one or more gestational sacs with or without an embryo or fetus in a multiple pregnancy documented by ultrasound.

sporadic cancer

- Cancer that occurs in individuals without a known inherited predisposition or family history.

Stem cells

- Cells that have the potential to multiply indefinitely and become many different cell types.

Sterility

- A permanent state of infertility.

Stillbirth

- A fetal death in the uterus after 20 weeks of pregnancy.
- The death of a fetus prior to the complete expulsion or extraction from its mother after 28 completed weeks of gestational age. The death is determined by the fact that, after such separation, the fetus does not breathe or show any other evidence of life, such as heartbeat, umbilical cord pulsation, or definite movement of voluntary muscles. Note: It includes deaths occurring during labor.

Stillbirth rate

- The number of stillbirths per 1000 total births (stillbirths plus live births).

Stimulated cycle

- An ART cycle where a woman receives oral or injected fertility drugs to stimulate her ovaries to develop follicles that contain mature eggs.

strong reciprocity

- A disposition to cooperate first, and to steadily increase cooperation in response to another player's willingness to cooperate, while also punishing defectors even at significant personal cost.

Subfertility

- A term that should be used interchangeably with infertility.

Substrate

- A chemical substance that takes part in a chemical reaction catalyzed by an enzyme.

sumLOD score

- The summation of all positive pedigree LOD scores at each point in the genome; also called summary logarithm of the odds score.

summary logarithm of the odds score

- The summation of all positive pedigree LOD scores at each point in the genome; also called sumLOD score.

surrogate

- See Gestational carrier.

surveillance

- The ongoing monitoring of individuals at increased risk of developing a disease or experiencing disease recurrence, through periodic testing and evaluation.

susceptibility gene mutation

- A genetic mutation that increases an individual's susceptibility to a disease; also called deleterious mutation, disease-causing mutation, pathogenic variant, and predisposing mutation.

synchronous cancer

- A new or second primary cancer that develops at the same time as the first primary cancer in the same or a different organ, with a similar molecular basis, and not considered metastatic relapse.

Syngamy

- The process during which the female and male pronuclei fuse.

TDR

- A third-degree relative: a first cousin, great-grandparent, great-aunt, great-uncle, great-niece, great-nephew, great-grandchild, or half-aunt/half-uncle.

telomere

- The end of a chromosome, composed of repetitive non-coding DNA sequences that protect the chromosome from damage and shorten with each cell division.

Telomere

- A repetitive DNA sequence at the end of a chromosome that protects it during cell division.

Teratozoospermia

- A reduced percentage of morphologically normal sperm in the ejaculate below the lower reference limits. When reporting results, the reference criteria should be specified.

TESA

- An acronym for testicular sperm aspiration.

TESE

- An acronym for testicular sperm extraction.

Testicular sperm aspiration/extraction (TESA/TESE)

- A surgical procedure involving one or more testicular biopsies or needle aspirations to obtain sperm for use in IVF and/or ICSI.

Testicular sperm extraction (TESE)

- Operative removal of testicular tissue in an attempt to collect living sperm for use in an IVF-ICSI procedure.

Thawing

- The process of raising the temperature of slow-frozen cell(s) from the storage temperature to room/ physiological temperature.

Thermodynamics

- The branch of physics dealing with the transformation of energy, especially of heat and other forms of energy.

third-degree relative

- A relative such as a first cousin, great-grandparent, great-aunt, great-uncle, great-niece, great-nephew, great-grandchild, or half-aunt/half-uncle; also called TDR.

throughput

- The amount of material or information processed through a system in a given time period, such as the number of tests performed.

Time to pregnancy (TTP)

- The time taken to establish a pregnancy, measured in months or in numbers of menstrual cycles.

Time-lapse imaging

- The photographic recording of microscope image sequences at regular intervals in ART, referring to gametes, zygotes, cleavage-stage embryos or blastocysts.

TNBC

- Triple-negative breast cancer; defined by the lack of expression of estrogen receptor, progesterone receptor, and HER2/neu; also called ER-negative PR-negative HER2/neu-negative breast cancer.

Total delivery rate with at least one live birth

- The estimated total number of deliveries with at least one live born baby resulting from one initiated or aspirated ART cycle including all fresh cycles and all frozen-thawed ART cycles. This rate is used when all of the embryos—fresh and/or frozen-thawed—have been used from one ART cycle. Note: The delivery of a singleton, twin, or other multiple pregnancy is registered as one delivery.
- The total number of deliveries with at least one live birth resulting from one initiated or aspirated ART cycle, including all cycles in which fresh and/or frozen embryos are transferred, including more than one delivery from one initiated or aspirated cycle if that occurs, until all embryos are used. Notes: The delivery of a singleton, twin or other multiple pregnancy is registered as one delivery. In the absence of complete data, the total delivery rate is often estimated.

Total fertility rate (TFR)

- The average number of live births per woman. It may be determined in retrospect, observed data (Cohort Total Fertility Rate, CTFR) or as an estimated average number (Period Total Fertility Rate, PTFR).

Total sperm count

- The calculated total number of sperm in the ejaculate (semen volume multiplied by the sperm concentration determined from an aliquot of semen).

Traditional gestational carrier

- A woman who donates her oocytes and is the gestational carrier for a pregnancy resulting from fertilization of her oocytes either through an ART procedure or insemination. This replaces the term 'traditional surrogate.'

Traditional surrogate

- A woman who carries a pregnancy intended for an infertile couple. The surrogate's egg is fertilized with sperm from the male partner of the infertile couple.

transcription

- The process of synthesizing messenger RNA (mRNA) from DNA.

Transcription

- The process of making a complementary sequence of the gene sequence in the genome, which is either used directly, as in case of Ribosomal RNAs (rRNAs) and Transfer RNAs (tRNAs), or is further processed into the messenger RNA, which is translated into protein. The process is catalyzed by the enzyme known as DNA-dependent RNA polymerase.

Transcription factors

- Proteins in eukaryotes that regulate the transcription of other genes by binding to regulatory sequences of the gene, interaction with one another and with the RNA polymerase.

Transduction

- In genetics, the transfer of genes by viruses from one organism to another.

Transfer RNA

- RNA molecules which transfer specific amino acids to the messenger RNA so that the polypeptide it encodes can be synthesized.

Transformation

- In genetics, the transfer of genes by one organism taking up DNA belonging to another organism of the same or different species.

Transgenic organism

- An organism created by genetic engineering, in which one or more foreign genes have been incorporated into its genome.

Translation

- The step in protein synthesis in which the messengerRNA directs the synthesis of a polypeptide of a particular amino-acid sequence by 'decoding' the genetic code.

translation

- The process of synthesizing a protein from the mRNA template by decoding its nucleotide sequence.

translocation

- A chromosomal abnormality in which a segment of one chromosome is transferred to another location, often on a different chromosome.

Transposon

- A mobile genetic element that can move around within a genome, causing mutations by changing its location.
- See Mobile genetic element.

Transvaginal ultrasound aspiration

- An ultrasound-guided technique for egg retrieval whereby a long, thin needle is passed through the vagina into the ovarian follicle and suction is applied to accomplish retrieval.

trichilemmoma

- A benign tumor arising from the outer cells of a hair follicle.

trinucleotide repeat

- A sequence of three nucleotides repeated consecutively in a segment of DNA. While some variation is normal, an excessive number can cause genetic disorders.

triple-negative breast cancer

- A breast cancer that lacks expression of estrogen receptor (ER), progesterone receptor (PR), and HER2/neu; also called TNBC.

trisomy

- The presence of an extra copy of a chromosome in some or all cells, resulting in three copies instead of two. For example, trisomy 21 causes Down syndrome.

Trisomy

- An abnormal number of chromosome copies in a cell characterized by the presence of three homologous chromosomes rather than the normal two. The majority of human embryos with trisomies are incompatible with life.

Trophectoderm

- Cells forming the outer layer of a blastocyst that have the potential to develop into the placenta and amniotic membranes.
- The outer layer of a blastocyst that forms the hollow ball in which the inner cell mass sits; it eventually forms the placenta and supporting tissues.

Tubal factor infertility

- A diagnostic category used when the woman's fallopian tubes are blocked or damaged, making it difficult for the egg to be fertilized or for an embryo to travel to the uterus.

Tubal pathology

- Tubal abnormality resulting in dysfunction of the Fallopian tube, including partial or total obstruction of one or both tubes (proximally, distally or combined), hydrosalpinx and/or peri-tubal and/or peri-ovarian adhesions affecting the normal ovum pick-up function. It usually occurs after pelvic inflammatory disease or pelvic surgery.

tumor DNA sequencing

- Sequencing of DNA from tumor cells to identify somatic mutations that occur during tumor development.

tumor suppressor gene

- A gene that regulates cell growth and prevents tumor formation; when mutated, it may lead to uncontrolled cell growth. Also called antioncogene.

Ultrasound

- A technology that uses high-frequency sound waves to form an image of internal organs on a monitor screen; used by fertility specialists to monitor the growth of ovarian follicles and to retrieve the eggs from the follicles and evaluate a pregnancy.

unaffected individual

- An individual who does not exhibit a particular phenotypic trait or disease.

unclassified variant

- A genetic variant for which the association with disease risk is unclear; also called a variant of uncertain significance or VUS.

Unexplained infertility

- A diagnostic category used when no cause of infertility is found in either the woman or the man.
- Infertility in couples with apparently normal ovarian function, Fallopian tubes, uterus, cervix and pelvis and with adequate coital frequency; and apparently normal testicular function, genito-urinary anatomy and a normal ejaculate. The potential for this diagnosis is dependent upon the methodologies used and/or those methodologies available.

uninformative result

- A negative test result in an individual where a clearly deleterious mutation has not been found, making the genetic risk status uncertain; also called inconclusive or indeterminate result.

Unisomy

- The condition in a cell resulting from loss of a single chromosome yielding a single copy of that particular chromosome rather than the normal two. The majority of unisomies in human embryos are incompatible with life.

Unstimulated cycle

- An ART cycle where the woman does not receive drugs to stimulate her ovaries to produce more follicles and eggs. Instead, follicles and eggs develop naturally.

Uterine factor infertility

- A structural or functional disorder of the uterus that results in reduced fertility.

Uterus (womb)

- The hollow, muscular female reproductive organ in the pelvis in which an embryo implants and grows during pregnancy. The lining of the uterus, called the endometrium, produces the monthly menstrual blood flow when there is no pregnancy.

Vagina

- The canal in the female that leads to the cervix, which leads to the uterus.

Vaginal insemination

- A procedure whereby semen, collected from a non-lubricated condom or similar method, is deposited into the vaginal cavity of a female. An intervention that can be self-administered by a woman attempting pregnancy.

Vanishing sac(s) or embryo(s)

- Spontaneous disappearance of one or more gestational sacs or embryos in an ongoing pregnancy, documented by ultrasound.

variable expressivity

- Variation in the range or severity of symptoms among individuals with the same genetic condition.

variant

- An alteration in the most common DNA sequence, which may be benign, pathogenic, or of uncertain significance.

variant of uncertain significance

- A genetic variant for which the association with disease risk is unclear; also called unclassified variant or variant of unknown significance.

variant of unknown significance

- A genetic variant for which the association with disease risk is unclear; also called unclassified variant or variant of uncertain significance.

Varicocele

- A venous enlargement in the testicular pampiniform plexus.

Varicocelectomy

- Procedure to occlude or remove part of the internal spermatic vein in situations in which it has expanded into a varicocele.

Vas deferens

- The two muscular tubes that carry sperm from the epididymis to the urethra.

Vasectomy

- Procedure to occlude the vas deferens. It is usually carried out bilaterally in order to secure sterilization.

Vector

- A carrier or transmitter, of genes or of disease. Artificial vectors are made in genetic engineering for multiplying and transferring genes into genomes.

Very low birth weight

- Birth weight less than 1,500 g. (More? Birth-Weight)
- Birth weight less than 1500 g.

Very preterm birth

- A live birth or stillbirth that takes place after 20 but before 32 completed weeks of gestational age.

Virulence

- Ability (of pathogens) to infect organisms and cause disease

Virus

- A parasitic genetic element enclosed in a protein coat that can replicate in cells, and form infectious particles, or remain dormant in the cells. Its genetic material can become integrated into the cell's genome to form provirus.

Viscosity

- The description of the relative fluidity of the semen.

Vitrification

- An ultra-rapid cryopreservation method that prevents ice formation within the suspension which is converted to a glass-like solid.
- An ultra-rapid cryopreservation procedure that prevents ice formation within a cell whose aqueous phase is converted to a glass-like solid.
- An ultra-rapid method of freezing eggs and embryos that may offer certain advantages compared with traditional types of cryopreservation.

Voluntary childlessness

- A condition describing a person who does not have or has not had a child wish and does not have any biologically, legally or societally-recognized children.

VUS

- A genetic variant for which the association with disease risk is unclear; also called unclassified variant, variant of uncertain significance, or variant of unknown significance.

Warming (cells)

- The process of raising the temperature of a vitrified cell or cells from the storage temperature to room/ physiological temperature.

weak reciprocity

- A disposition to conditionally cooperate.

WES

- Whole exome sequencing; a laboratory process that sequences the protein-coding regions of the genome (approximately 1% of total DNA). Also called WES or WXS.

WGS

- Whole genome sequencing; a laboratory process that sequences nearly the entire genome, including both coding and non-coding regions. Also called WGS.

whole exome sequencing

- A laboratory process that sequences primarily the protein-coding regions of the genome (about 1% of total DNA). Also called WES or WXS.

whole genome sequencing

- A laboratory process that sequences nearly the entire DNA sequence of an individual's genome, including non-coding regions. Also called WGS.

WXS

- A laboratory process that sequences primarily the protein-coding regions of the genome (about 1% of total DNA). Also called WES or whole exome sequencing.

X-linked dominant inheritance

- A mode of inheritance for conditions caused by mutations on the X chromosome where a single copy of the mutation causes disease in both males and females. In some cases, affected males may not survive.

X-linked recessive inheritance

- A mode of inheritance for conditions caused by mutations on the X chromosome where males are typically affected because they have only one X chromosome, while females are usually carriers if they have one mutated allele.

Y-chromosome microdeletions

- Missing segments of the genetic material on the Y-chromosome that are associated with abnormal spermatogenesis.

ZIFT

- See Zygote intrafallopian transfer.

ZIFT (zygote intrafallopian transfer)

- An ART procedure where eggs are collected from a woman's ovary and fertilized outside her body. A laparoscope is then used to place the resulting zygote (fertilized egg before it begins to divide) into the woman's fallopian tube through a small incision in her abdomen.

Zona pellucida

- The egg's outer layer that a sperm must penetrate in order to fertilize the egg.
- The glycoprotein coat surrounding the oocyte.

Zygote

- A diploid cell resulting from the fertilization of an oocyte by a spermatozoon, which subsequently divides to form an embryo.
- A fertilized egg before cell division (cleavage) begins.
- A fertilized egg. The DNA from the sperm and egg merge as the zygote divides into a two-cell embryo.
- A single cell resulting from fertilization of a mature oocyte by a spermatozoon and before completion of the first mitotic division.

Zygote intrafallopian transfer

- (ZIFT) A procedure in which zygote(s) is/are transferred into the fallopian tube.

Zygote intrafallopian transfer (ZIFT)

- An ART procedure in which one or more zygotes is transferred into the Fallopian tube.

Zygote intrafallopian tube transfer (ZIFT)

- An egg is fertilized in the laboratory and the zygote is transferred to the fallopian tube before cell division takes place. Eggs are retrieved and fertilized on one day and the embryo is transferred the following day.

Part VIII

Appendix

21 Frequently Asked Questions

21.1 Why Is This On GitHub?

- …will have Python code for analyzing Google Scholar and Trends signals on genetic and reproductive technologies.
- …involves versions, specifically for prize sequences and groups of questions that are released.
- …involves a website hosted via GitHub Pages.
- …operates by open source principles (open development).

22 Trends

23 Additional Remarks

See here.

See here.