Genetic Screening Key Terms

Aneuploidy – Having extra or missing chromosomes, resulting in a change in the number of chromosomes in the cell

Cell-free DNA (cfDNA) – Small pieces of DNA that often circulate in the blood stream during natural cellular processes

Chromosomal abnormality – A change in DNA that affects the structure or number of chromosomes, and can sometimes affect the cell's ability to function or survive

Chromosome – A DNA molecule that is tightly packed with the genetic material for each cell; every cell should contain 23 pairs of chromosomes, 46 in total

Down syndrome – A condition that results from the body's cells having a full or partial extra copy of chromosome 21

Edwards syndrome – A condition that results from the body's cells having a full or partial extra copy of chromosome 18

False-negative – When the outcome of a screen incorrectly indicates that the result is negative, when the real result is positive

False-positive – When the outcome of a screen incorrectly indicates that the result is positive, when the real result is negative

Fetus – An unborn baby, from eight weeks after conception until birth

Genetic counselor – A healthcare professional who has received training in medical genetics and counseling. Genetic counselors help individuals and families assess the risk of genetic conditions, understand genetic screening results, and determine best approaches for care.

Genetic screening – A test used to identify changes in DNA structure or sequence in a fetus or in an adult that can be passed on to their future children

Informed consent – A principle in medical ethics and law, in which patients are provided with sufficient and understandable information, including possible risks and benefits, and the freedom of choice in advance of making a decision about their medical care

Noninvasive prenatal screening (NIPS)– A type of genetic screening that is used to assess the risk of chromosomal abnormalities in fetal DNA; also known as noninvasive prenatal testing (NIPT) or cell-free DNA (cfDNA) testing

Microdeletion – A chromosomal abnormality where a tiny piece of the chromosome is missing

Patau syndrome – A condition that results from the body's cells having a full or partial extra copy of chromosome 13

Placenta – An organ that develops and attaches to the uterus during pregnancy to facilitate the exchange of nutrients, oxygen, and waste between mother and baby

Trisomy – Having an extra (third) copy of a certain chromosome, instead of two

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