



National Coordinating Center  
for the Regional Genetics Networks

The National Coordinating Center for the  
Regional Genetics Networks  
国家区域遗传学  
网络协调中心

## Glossary of Prenatal Genetics Terms 产前遗传学术语表

English – Chinese  
英文 – 中文

2022 Edition

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## Acknowledgments

### Mission of the NCC

The National Coordinating Center for the Regional Genetics Networks (NCC) has been funded by the Health Resources and Services Administration (HRSA) since 2004. Our mission is to improve access to genetics for underserved populations. This mission is accomplished through different programs, many of which are educational tools and resources for non-genetics professionals. Learn more at <https://nccrcg.org>.

### NCC Funding Acknowledgment

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## 致谢

### NCC 的使命

自 2004 年以来，国家区域遗传学网络协调中心 (NCC) 一直由卫生资源和服务管理局 (HRSA) 资助。我们的使命是改善服务水平不足的人群了解遗传学的机会。这一使命是通过不同的计划来完成的，其中许多是针对非遗传学专业人士的教育工具和资源。访问 <https://nccrcg.org> 可了解更多信息。

### NCC 资助致谢

该项目由美国卫生与公众服务部 (HHS) 的卫生资源和服务管理局 (HRSA) 根据合作协议 #UH9MC30770 于 2020 年 6 月至 2024 年 5 月期间以每年 800,000 美元的金额提供支持。这些信息或内容和结论出自作者，不应被理解为 HHS 的 HRSA 或美国政府的官方立场或政策，也不应被推断为其的任何赞许。

## Additional Acknowledgments

### Cynthia E. Roat, MPH

Cindy Roat is an international consultant in language access in health care and patient navigation. She provides training and consulting for health care interpreters, providers, administrators, and patient navigators. Cindy compiled and edited this glossary for the NCC.

### The Cross Cultural Health Care Program

The mission of The Cross Cultural Health Care Program (CCHCP) is to serve as a bridge between communities and health care institutions to advance access to quality health care that is culturally and linguistically appropriate. CCHCP provides resources, such as bilingual medical glossaries, as well as medical interpreter, patient navigator, and cultural competence training for individuals and institutions with the goal of systems change. Learn more at: <https://xculture.org>

CCHCP is supporting and coordinating the translation process of this glossary and is providing the glossary free of charge in its online store: <https://xculture.org/store/>

## 特别致谢

### Cynthia E. Roat, MPH

Cindy Roat 是一位医疗保健和患者导航语言访问方面的国际顾问。她为医疗保健口译员、提供者、管理人员和患者导航员提供培训和咨询。Cindy 为 NCC 编译和编辑了本术语表。

### The Cross Cultural Health Care Program

跨文化医疗保健计划 (CCHCP) 的使命是充当社区和医疗保健机构之间的桥梁，以促进获得在文化和语言上适当的优质医疗保健。CCHCP 以系统变革为目标，为个人和机构提供双语医学词汇表、医学口译员、患者导航员和文化能力培训等资源。访问 <https://nccrcg.org> 可了解更多信息。

CCHCP 支持和协调本术语表的翻译过程，并在其在线商店免费提供术语表：  
<https://xculture.org/store/>

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## Preface

The seven Regional Genetics Networks (RGNs), the National Genetics Education and Family Support Center (Family Center), and the National Coordinating Center for the Regional Genetics Networks (NCC) mission is to improve access to quality genetic services for medically underserved populations. This bilingual glossary of terms related to Prenatal Genetics is provided to help enable access to prenatal medical services to limited English proficiency populations.

## 前言

七个区域遗传学网络 (RGN)、国家遗传学教育和家庭支持中心（家庭中心）和国家区域遗传学网络协调中心 (NCC) 的使命是改善医疗服务水平不足人群获得优质遗传学服务的机会。提供与产前遗传学相关的双语术语表，以帮助英语能力有限的人群获得产前医疗服务。

## How to Use the Glossary

This glossary is organized alphabetically in English. The first column shows the term in English. The second column has the definition of the term in English. The third column has the translation of the term.

At the end of the translations of the prenatal genetics terms are:

- A glossary of translations for family relationships terms. It is very important for genetic counselors to know if a relative is a blood relative or a relative by marriage.
- Examples of analogies used in genetics. Genetic counselors often use analogies to explain complex ideas in genetics. An analogy explains a new concept by comparing it to something more common.

## 如何使用该术语表

Glossary of Prenatal Genetics Terms – Chinese  
产前遗传学术语表

本术语表按英文字母顺序排列。第一列显示英文术语。第二列提供术语的英文定义。第三列是术语的翻译。

产前遗传学术语翻译的后面是：

- 家庭关系术语的翻译术语表。对于遗传咨询师来说，了解亲属是血缘亲属还是婚姻亲属非常重要。
- 遗传学中使用的类比示例。遗传咨询师经常使用类比来解释遗传学中的复杂想法。类比通过将一个新概念与更常见的事物进行比较来解释它。

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**Glossary of Terms****术语表****English – Chinese****英文 – 中文**

(n) – noun      (v) – verb      (adj) – adjective      (adv) – adverb  
 (n) – 名词      (v) – 动词      (adj) – 形容词      (adv) – 副词

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Abnormal (adj)</b>	Different from what is considered normal, average, or expected, e.g., a gene sequence that is different than that found in most people. 不同于被认为是正常的、平均的或预期的，例如，与在大多数人中发现的不同的基因序列。	異常的	异常的
<b>Abortion (n)</b>	The deliberate ending of a pregnancy by the removal of an embryo or fetus from the womb. 通过从子宫中取出胚胎或胎儿来故意终止妊娠。 Technically, this is called an “induced abortion” in order to distinguish it from a “spontaneous abortion” which is also called a miscarriage or stillbirth. Also called a “termination of pregnancy” or just “termination.”从技术上讲，这被称为“人工流产”，以便与“自然流产”区分开来，“自然流产”也称为流产或死产。也称为“终止妊娠”或仅称为“终止。”	墮胎	堕胎
<b>Adoption (n)</b>	The process of legally taking someone else’s child into your family to raise as your own child.  合法地将他人的孩子带入您的家庭，作为您自己的孩子抚养的过程。	領養	收养
<b>Amniocentesis (n)</b>	A procedure by which a sample of amniotic fluid is withdrawn from the amniotic sac. This is usually done by inserting a long needle through the abdominal and uterine walls with the guidance of ultrasound.  从羊膜囊中抽取羊水样本的程序。这通常是在超声引导下通过腹壁和子宫壁插入一根长针来完成的。	羊膜穿刺術	羊膜穿刺术

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Amniotic fluid (n)</b>	<p>The liquid that surrounds a fetus as it develops in the uterus. This liquid contains skin cells that have sloughed off the fetus as well as other fetal cells.</p> <p>胎儿在子宫内发育时包围胎儿的液体。这种液体含有从胎儿脱落的皮肤细胞以及其他胎儿细胞。</p>	羊水	羊水
<b>Anencephaly (n)</b>	<p>A neural tube defect that results in insufficient brain growth in a fetus. Babies with anencephaly usually die soon after birth or are stillborn.</p> <p>导致胎儿大脑发育不足的神经管缺陷。患有无脑畸形的婴儿通常在出生后不久就会死亡或死产。</p>	無腦畸形	无脑畸形
<b>Benign (adj)</b>	<p>Something that does not threaten health or life. May refer to a type of change to the DNA that does not create health consequences.</p> <p>不会威胁健康或生命的物质。可能是指一种不会造成健康后果的 DNA 变异。</p> <p>E.g., when discussing cancer, “benign” means “not cancerous.”</p> <p>例如，在讨论癌症时，“良性”的意思是“没有癌变。”</p>	良性的	良性的
<b>Birth canal (n)</b>	<p>The passageway from the uterus through the cervix, the vagina, and the vulva through which a baby passes during the birth process.</p> <p>胎儿在分娩过程中从子宫经过子宫颈、阴道和外阴的通道。</p>	產道	产道
<b>Birth defect (n)</b>	<p>A problem or physical difference with how the body works that is present at birth. Birth defects can be caused by genetic abnormalities, environmental influences, random chance, or by circumstances related to the birth process. Also called “congenital malformation” or “congenital anomaly.”</p> <p>出生时出现的身体工作方式的问题或身体差异。出生缺陷可能由遗传异常、环境影响、随机机会或与出生过程相关的情况引起。也称为“先天性畸形”或“先天性异常”。</p>	先天性缺陷	先天性缺陷

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Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Blood test (n)</b>	A test in which blood is drawn (usually from the patient's arm) and sent to a laboratory for analysis.  抽血（通常从患者的手臂）并送到实验室进行分析的检测。  In genetics, blood tests may provide cells for genetic sequencing, or blood samples may be used to assess things like protein or hormone levels.  在遗传学中，验血可以为基因测序提供细胞，或者血液样本可用于评估蛋白质或激素水平等。	驗血	验血
<b>Buccal swab (n)</b>	A way to collect DNA from the cells on the inside of a person's cheek.  一种从人脸颊内侧的细胞中收集 DNA 的方法。	口腔黏膜測試	口腔黏膜检测
<b>Carrier(n)</b>	A person who has a genetic mutation in one of their two copies of a particular gene that is associated with a genetic condition. The other copy of the gene does not have a mutation, so that person usually does not have any medical issues related to the gene change.  在与遗传状况相关的特定基因的两个拷贝之一中具有基因突变的人。该基因的另一个拷贝没有突变，因此该人通常没有任何与基因变化相关的医疗问题。	基因攜帶者	携带者
<b>Cell (n)</b>	The smallest (microscopic) functional unit of living organisms. All living things are composed of one or more cells. Within each cell are the structures needed for the cell to process energy, dispose of waste, reproduce, and perform specialized functions.  生物体的最小（微观）功能单位。所有生物都是由一个或多个细胞组成的。每个细胞内都有细胞处理能量、处理废物、繁殖和执行特殊功能所需的结构。	細胞	细胞
<b>Cervix (n)</b>	The narrow lower part of the uterus than connects to the vagina.  子宫与阴道相连的较窄的下部。	子宮頸	子宫颈

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Chorionic Villus Sampling (CVS) (n)</b>	A procedure during pregnancy in which a sample of cells from the placenta is removed to check for possible genetic abnormalities. 怀孕期间从胎盘中取出细胞样本以检查可能的遗传异常的程序。	绒毛膜绒毛取样 (CVS)	绒毛膜绒毛取样 (CVS)
<b>Chromosome (n)</b>	Thread-like structures located inside the nucleus of cells. In humans, there are 23 pairs of chromosomes, for a total of 46 chromosomes. Each chromosome is made of a long strand of DNA, which carries genetic information. 位于细胞核内的线状结构。在人类中，有 23 对染色体，总共有 46 条染色体。每条染色体由长链 DNA 组成，携带遗传信息。	染色體	染色体
<b>Chromosome test / Karyotype (n)</b>	A test that looks at the number and types of chromosomes in a cell. 一项检查细胞中染色体数量和类型的检测。	染色體檢測/核型	染色体检测/核型
<b>Cleft lip (n)</b>	A congenital malformation that occurs when the upper lip does not form properly, causing an opening in the upper lip that can extend to the nose. This can occur together with a cleft palate or on its own. 上唇不能正常形成时发生的先天性畸形，导致上唇开口可延伸至鼻子。这可以与腭裂一起发生或单独发生。	兔唇	唇裂
<b>Cleft palate (n)</b>	A congenital malformation where the roof of the mouth (palate) does not form properly, resulting in an opening into the nasal cavity. This can occur together with a cleft lip or on its own. 口腔顶部（上颚）不能正常形成的先天性畸形，导致鼻腔开口。这可能与唇裂一起发生或单独发生。	顎裂	腭裂
<b>Clinically significant (adj)</b>	If a test result is “clinically significant,” the result indicates a medical problem that can impact a person’s life. 如果检测结果具有“临床意义”，则该结果表明存在可能影响一个人生活的医疗问题。	有臨床意義的 / 有臨床重要性的	有临床意义的
<b>Condition (e.g., “genetic condition”) (n)</b>	A long-term medical health issue. 一个长期的医疗健康问题。	病症（例如 「遺傳病」）	疾病（例如“遗 传病”）
<b>Consanguinity (n)</b>	When parents are blood relatives to each other. 当父母彼此是血亲时。	血緣	血缘

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Cystic fibrosis (n)</b>	<p>A progressive genetic condition that affects the exocrine glands (the glands that make sweat and digestive juices) and causes the production of thick, sticky mucus. This mucus blocks the pancreatic duct, the intestines, and the lungs, leading to persistent respiratory infections.</p> <p>一种渐进性遗传病，会影响外分泌腺（产生汗液和消化液的腺体）并导致产生粘稠的粘液。这种粘液会阻塞胰管、肠道和肺部，导致持续性呼吸道感染。</p>	囊性纖維化	囊性纤维化
<b>Deleterious mutation/ Disease-causing mutation (n)</b>	<p>A change in a person's DNA that may cause a medical condition.</p> <p>一个人的DNA变化，其可能会导致某种疾病。</p>	有害突變／ 致病性突變	有害突变/ 致病突变
<b>Deletion (n)</b>	<p>Having a section of genetic information (DNA) missing.</p> <p>缺少一段遗传信息 (DNA)。</p>	缺失	缺失
<b>Diagnostic test (n)</b>	<p>A medical test that determines whether a patient has a particular medical problem. Diagnostic tests are often used when providers have a specific reason to believe that the medical problem may be present. Compare this to screening tests, which may be given routinely, even if the provider has no reason to believe the patient has a specific problem. Screening tests often only report whether a patient is at <u>an increased risk</u> for the medical problem in question, whereas diagnostic tests report whether the problem is actually present.</p> <p>确定患者是否有特定医疗问题的医学检测。当提供者有特定理由相信可能存在医疗问题时，通常会使用诊断检测。将此与筛查检测进行比较，即使提供者没有理由相信患者有某种问题，也可以常规进行筛查检测。筛查检测通常只报告患者是否存在相关医疗问题的<u>风险增加</u>，而诊断检测则报告问题是否实际存在。</p>	診斷性測試	诊断检测

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Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>DNA (Deoxyribonucleic Acid) (n)</b>	<p>The material that carries the genetic information of a cell. It provides the instructions used in the development, functioning and reproduction of the organism of which it is a part.</p> <p>携带细胞遗传信息的物质。它提供了在其所属的有机体的发育、功能和繁殖中使用的说明。</p> <p>If you could stretch out the DNA of a chromosome and look at it through a microscope, it would look like a long ladder that is twisted into a spiral. The ‘sides’ of the ladder are made up of alternating phosphate and sugar groups. The ‘rungs’ are various combinations of two nitrogen bases: Adenine-Thymine and Cytosine-Guanine.</p> <p>如果您可以拉伸染色体的 DNA 并通过显微镜观察它，它看起来就像一个扭曲成螺旋形的梯子。梯子的“边”由交替的磷酸盐和糖基组成。“梯级”是两种氮碱基的各种组合：腺嘌呤-胸腺嘧啶和胞嘧啶-鸟嘌呤。</p> <p>Individual sections of DNA that code for specific traits/functions are called genes.</p> <p>编码特定特征/功能的 DNA 的各个部分称为基因。</p>	DNA (脱氧核糖核酸)	DNA (脱氧核糖核酸)
<b>DNA marker (also called a “Genetic marker”) (n)</b>	<p>A readily recognizable genetic trait, gene, or DNA segment.</p> <p>易于识别的遗传特征、基因或 DNA 片段。</p>	脱氧核糖核酸 标记（也稱為 遺傳標記）	脱氧核糖核酸 标记（也称为 “遗传标记”）
<b>DNA mutation (n)</b>	<p>A change in the typical sequence of the chemicals that make up the DNA, like the change in the order of letters in a word.</p> <p>组成 DNA 的化学物质的典型序列发生变化，就像单词中字母顺序的变化。</p> <p>Mutations or variants are often compared to misspelled words because chemicals that make up the DNA sequence are not in the expected order.</p> <p>突变或变异通常与拼写错误的单词进行比较，因为构成 DNA 序列的化学物质不是按预期顺序排列的。</p>	脱氧核糖核酸 突變	脱氧核糖核酸 突变
<b>DNA sequence (n)</b>	<p>The exact arrangement of the chemicals that make up a section of DNA.</p> <p>构成 DNA 部分的化学物质的确切排列。</p>	脱氧核糖核酸 序列	脱氧核糖核酸 序列

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>DNA sequencing (n)</b>	<p>The laboratory technique used to determine the exact arrangement of the chemicals that make up a section of DNA. This is one type of genetic testing.</p> <p>用于确定组成DNA部分的化学物质的确切排列的实验室技术。这是基因检测的一种。</p>	脱氧核糖核酸定序	脱氧核糖核酸测序
<b>Dominant (adj)</b>	<p>A genetic trait in which one copy of the gene is sufficient for a trait to be expressed. In a dominant genetic condition, if one copy of the gene has a mutation, the person will be affected with the condition.</p> <p>一种遗传性状，其中一个基因拷贝就足以表达一种性状。在显性遗传条件下，如果基因的一个拷贝发生突变，则个人将受到该条件的影响。</p>	顯性的/顯著的	显性的
<b>Donor egg or sperm (n)</b>	<p>An egg (singular) or sperm (plural) donated by one person to be joined under laboratory conditions and implanted in a woman's uterus. The donor egg or sperm may come from the woman or man who will raise any resulting child, or they may come from a third party.</p> <p>由一个人捐赠的卵子（单数）或精子（复数）在实验室条件下结合并植入女性子宫。捐赠的卵子或精子可能来自将抚养任何因此而得孩子的女性或男性，也可能来自第三方。</p>	捐赠卵子或精子	捐赠卵子或精子
<b>Down syndrome (Trisomy 21) (n)</b>	<p>A genetic condition in which there are three copies of chromosome number 21 instead of two.</p> <p>一种遗传病，其中21号染色体存在三个拷贝，而不是两个。</p> <p>Down syndrome causes a distinct facial appearance, intellectual disability, developmental delays, and may be associated with thyroid or heart disease.</p> <p>唐氏综合症会导致明显的面部外观、智力障碍、发育迟缓，并可能与甲状腺或心脏病有关。</p>	唐氏綜合症 (三染色體21 症)	唐氏综合征 (三染色体 21 症)
<b>Duplication (n)</b>	<p>Having an extra section of genetic information (DNA). A duplication occurs when part of a chromosome is copied abnormally, resulting in extra genetic material.</p> <p>有一段额外的遗传信息(DNA)。当染色体的一部分被异常拷贝时，会发生复制，从而导致额外的遗传物质。</p>	複製	复制

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Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Edwards syndrome (Trisomy 18) (n)</b>	A genetic condition in which there are three copies of chromosome number 18 instead of two. 一种遗传病，其中 18 号染色体存在三个拷贝，而不是两个。  Trisomy 18 is a very severe condition that causes problems with the brain, the heart, the kidneys, and the digestive tract. Most children affected by trisomy 18 die before or soon after birth. 三染色体 18 症是一种非常严重的疾病，会导致大脑、心脏、肾脏和消化道出现问题。大多数受三染色体 18 症影响的儿童在出生前或出生后不久就会死亡。	愛德華氏症 (三染色體18 症)	爱德华氏症 (三染色体 18 症)
<b>Egg(n)</b>	The reproductive cells of a woman. When fertilized by sperm, the egg will grow into an embryo. Also called an “ovum,” plural “ova.” 女性的生殖细胞。当受精后，卵子会长成胚胎。也称为“ovum”（卵细胞），复数为“ova。”	卵子	卵子
<b>Embryo (n)</b>	An unborn mammal, between conception and 8 weeks of gestation. 一种未出生的哺乳动物，介于受孕和妊娠 8 周之间。	胚胎	胚胎
<b>Fallopian tube (n)</b>	The tube that connects an ovary to the uterus. 连接卵巢和子宫的管子。	输卵管	输卵管
<b>False negative (n)</b>	A test result that finds no evidence of a condition when the condition actually does exist. For example, a false negative on a pregnancy test finds that the woman is not pregnant when, in fact, she is pregnant. 当条件确实存在时，没有发现条件证据的检测结果。例如，妊娠检测的假阴性发现该女性实际上没有怀孕，而实际上她怀孕了。	假陰性	假阴性
<b>False positive (n)</b>	A test result that finds evidence of a condition when the condition does NOT actually exist. For example, a false positive on a pregnancy test finds that the woman is pregnant when, in fact, she is not. 当条件实际上不存在时找到条件证据的检测结果。例如，妊娠检测的假阳性发现该妇女怀孕了，而实际上她没有怀孕。	假陽性	假阳性

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<b>Familial (adj)</b>	Belonging to a family, e.g., a familial trait is a trait that is shared among family members and may be due to genetic or environmental factors or both. 属于一个家族，例如，家族特征是家族成员共有的特征，可能是由于遗传或环境因素或两者兼而有之。	家族性的	家族性的
<b>Family history (n)</b>	The medical history of the members of a biological family. 生物家族成员的病史。	家族史	家族史
<b>Fertilization (n)</b>	The joining of an egg and sperm to create the first cell that will develop into an embryo, then fetus, then baby. 卵子和精子的结合产生第一个将发育成胚胎的细胞，然后是胎儿，然后是婴儿。	受精	受精
<b>Fetal surgery (n)</b>	Surgery conducted on a fetus while it is still in the uterus. 对仍在子宫内的胎儿进行手术。	胎兒手術	胎儿手术
<b>Fetus (n)</b>	An unborn mammal, between 8 weeks of gestation to birth. 一种未出生的哺乳动物，介于受孕和妊娠 8 周之间。	胎兒	胎儿
<b>First trimester screening (n)</b>	A blood test and ultrasound conducted at 10-13 weeks of pregnancy to screen for Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), and certain other fetal problems. 在怀孕 10-13 周进行血液检查和超声波检查，以筛查唐氏综合症（三染色体 21 症）、爱德华氏症（三染色体 18 症）和某些其他胎儿问题。	懷孕第一期篩檢	孕早期筛查

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Flip a coin (v)</b>	<p>A random decision-making tool used in the U.S. While a coin is flipped into the air and caught, a person predicts whether it will fall with the “heads” side up or the “tails” side up. If the coin falls as predicted, the person “wins.” This expression is often used as a metaphor for any outcome that is random and has two possible outcomes, and to describe a situation in which each outcome is as likely as the other.</p> <p>在美国使用的一种随机决策工具。当一枚硬币被抛向空中并被抓住时，人们预测它落下时是“正面”朝上还是“反面”朝上。如果硬币如预期般落下，则此人“获胜。”这个表达经常被用作任何随机结果的隐喻，并且有两种可能的结果，并描述一种情况，在这种情况下，每个结果都与另一个结果一样可能。</p>	拋幣而定（廣東話俗語「擲銀仔」）	掷硬币
<b>Fragile X syndrome (n)</b>	<p>A genetic condition that affects the FMR1 gene so that it does not function properly. This syndrome causes intellectual disability, behavioral and learning challenges and various physical characteristics that are not life threatening.</p> <p>影响 FMR1 基因使其无法正常工作的遗传病。这种综合征会导致智力障碍、行为和学习挑战以及各种不危及生命的身体特征。</p>	染色體脆弱症	脆性 X 综合征
<b>Fragment (n)</b>	<p>A small piece; an incomplete part of a whole.</p> <p>一小块；整体的不完整部分。</p>	碎片	片段
<b>Gene (n)</b>	<p>A specific sequence of DNA that codes for one or many functions within the cell and body.</p> <p>编码细胞和身体内的一种或多种功能的特定 DNA 序列。</p>	基因	基因
<b>General population (n)</b>	<p>“Most people.”</p> <p>“大多数人。”</p> <p>For example, if you have the same risk of getting cancer as the general population, that means that you have the same chance of getting cancer as everyone else. This is in contrast to a “high risk population” who has a greater chance of getting cancer than everyone else.</p> <p>例如，如果你患癌症的风险与普通人群相同，这意味着你患癌症的机会与其他人相同。这与“高危人群”形成鲜明对比，他们患癌症的几率比其他人都高。</p>	一般人口	一般人群

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<b>Generation (n)</b>	<p>The people who constitute a single step in a line of descent from an ancestor; a group of people born and living more or less at the same time.          从祖先的血统中构成一步的人；一群或多或少同时出生和生活的人。</p> <p>Example: You, your brothers, and sisters, all your spouses and your cousins are in the same generation. Your parents, your aunts, and uncles and all their spouses form a previous generation. Your grandparents, their siblings, and spouses from an even earlier generation. Your children and nieces and nephews form a later generation.          例如：你、你的兄弟姐妹、你所有的配偶和你的堂兄弟姐妹都是同一代人。你的父母、姑姑、叔叔和他们所有的配偶构成了上一代。你的祖父母、他们的兄弟姐妹和配偶是更早一代的。你的孩子、侄女和侄子组成了后代。</p>	一代人 / 同一代人	一代人
<b>Genetics (adj)</b>	<p>The science of how an organism's genes interact with the environment to produce certain traits.          有机体基因如何与环境相互作用以产生某些特征的科学。</p>	遺傳學的	遗传学的
<b>Genetic counseling (n)</b>	<p>A discussion with a medical professional with expertise in genetics about the basic concepts of genetics, genetic conditions, the chances of being affected by a genetic condition or having a child with a genetic condition, and genetic testing and treatment.          与具有遗传学专业知识的医学专家讨论遗传学的基本概念、遗传病、受遗传病影响或生下患有遗传病儿童的机会以及基因检测和治疗。</p>	遺傳諮詢	遗传咨询
<b>Genetic counseling intern (n)</b>	<p>A genetic counseling student who has not yet completed his/her academic studies and is now practicing under the supervision of a more experienced counselor in preparation for providing genetic counseling services independently after obtaining his/her graduate degree.          尚未完成学业的遗传咨询学生，目前正在一位更有经验的咨询师的指导下实习，准备在获得研究生学位后独立提供遗传咨询服务。</p>	遺傳諮詢實習生	遗传咨询实习生

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Genetic counselor (n)</b>	<p>A healthcare professional with a specialized graduate degree who works with people who have concerns about genetic conditions in their family. Genetic counselors provide information about genetic conditions, help patients understand their chances of being affected by a genetic condition or having a child with a genetic condition, and help them make informed decisions about testing and treatment. Genetic counselors also provide emotional support to patients and families.</p> <p>服务于担心家庭遗传病的人的拥有专业研究生学位的医疗保健专业人士。遗传咨询师提供有关遗传病的信息，帮助患者了解他们受到遗传病影响或生下患有遗传病孩子的机会，并帮助他们就检测和治疗做出明智的决定。遗传咨询师还为患者和家属提供情感支持。</p>	遺傳諮詢師/遺傳顧問	遗传咨询师
<b>Genetic discrimination (n)</b>	<p>Occurs when people are treated differently by their employer or insurance company due to their genetic makeup. There are federal and state laws that help protect against genetic discrimination.</p> <p>当人们因其基因构成而受到雇主或保险公司的不同对待时，就会发生这种情况。制定联邦和州法律有助于防止基因歧视。</p>	基因歧視	基因歧视
<b>Genetic factors (n)</b>	<p>Specific aspects of a person's genetic make-up that influence that person's health and development.</p> <p>一个人的基因构成的特定方面会影响该人的健康和发育。</p>	遺傳因素	遗传因素
<b>Genetic information (n)</b>	<p>The instructions encoded in DNA, which tells every cell in a body how to grow, what to do and how to reproduce.</p> <p>DNA 中的编码指令，解释身体中的每个细胞如何生长、做什么以及如何繁殖。</p>	遺傳資訊	遗传信息
<b>Genetic material (n)</b>	<p>All the parts of a cell that carry genetic information. Genetic material could include genes, parts of genes, a group of genes, a DNA molecule, a fragment of DNA, a group of DNA molecules, or the entire set of genetic instructions.</p> <p>携带遗传信息的细胞的所有部分。遗传物质可以包括基因、部分基因、一组基因、一个DNA分子、一个DNA片段、一组DNA分子或整个遗传指令集。</p>	遺傳物質	遗传物质

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<b>Genetic predisposition (n)</b>	An increased chance of a person developing a certain trait or disease based on that person's particular genetic makeup. 根据一个人的特定基因构成，该人发展某种特征或疾病的机会增加。	遺傳傾向性	遗传倾向性
<b>Genetic test (n)</b>	A laboratory test designed to determine if a person has a change to their DNA. 一种实验室检测，旨在确定一个人的 DNA 是否发生了变化。	基因檢測	基因检测
<b>Genetic trait (n)</b>	A characteristic within a family that is passed down from parent to child through their DNA. 一个家庭的特征，通过他们的 DNA 从父母传给孩子。	遺傳特徵	遗传特征
<b>Genetic variant (n)</b>	A change from the typical DNA sequences. A genetic variant can be benign, deleterious or of uncertain significance. Also called "mutation," although "genetic variant" is becoming the more common usage. 从典型 DNA 序列的变化。遗传变异可能是良性的、有害的或意义不明的。也称为“突变”，尽管“遗传变异”正在成为更常见的用法。	遺傳變異	遗传变异
<b>Geneticist(n)</b>	A doctor or scientist who studies genetics. 研究遗传学的医生或科学家。	遺傳學家	遗传学家
<b>Hereditary (adj)</b>	Passed down from parent to child. 从父母传给孩子。	遺傳性的	遗传性的
<b>Hereditary material (n)</b>	Genetic material that is passed down from parent to child. 从父母传给孩子的遗传物质。	遺傳物質	遗传物质
<b>In Vitro Fertilization (IVF) (n)</b>	The fertilization of an egg by a sperm outside of a woman's body. 精子使女性体外的卵子受精。 The process involves extracting eggs from a woman's ovaries, collecting sperm from a man, and combining a sperm and egg in a laboratory dish. The resulting fertilized egg is usually then implanted in a woman's uterus so that it can develop into a baby. 该过程包括从女性卵巢中提取卵子，从男性身上收集精子，并在实验室培养皿中将精子和卵子结合起来。产生的受精卵通常随后被植入女性的子宫中，这样它就可以发育成婴儿。	體外人工受精 (IVF)	体外人工受精 (IVF)

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<b>Infertility (n)</b>	The inability to have children. 不能生孩子。	不孕症	不孕症
<b>Informed consent (n)</b>	The process of agreeing to a procedure or course of treatment after understanding what the procedure/treatment entails, the potential risks and benefits associated with it, and the other options available. 在了解程序/治疗所涉及的内容、与之相关的潜在风险和益处以及其他可用方案后，同意该程序或治疗过程的过程。	知情同意書	知情同意书
<b>Inheritance pattern (n)</b>	The manner in which a particular genetic trait or disorder is passed from a parent to a child, e.g., autosomal dominant or recessive, X-linked dominant or recessive, or multifactorial. 特定遗传特征或疾病从父母传给孩子的方式，例如常染色体显性或隐性遗传、X 连锁显性或隐性遗传或多因素遗传。	遺傳模式	遗传模式
<b>Inherited (adj)</b>	Passed down from parent to child. 从父母传给孩子。	遺傳的	遗传的
<b>Insertion (n)</b>	Having an extra segment of DNA added in at a place where it is not usually found. 在通常不被发现的地方添加了一段额外的DNA。	插入	插入
<b>Intellectual disability (n)</b>	A condition, varying in severity, in which a person has significant impairments in mental abilities, social skills, and core functions of daily living compared to others their age. 一种严重程度不同的病症，与同龄人相比，一个人在心智能力、社交技能和日常生活的核心功能方面存在显著障碍。	智力障礙	智力障碍
<b>Klinefelter's syndrome (47, XXY) (n)</b>	A genetic condition in which a male has two copies of the X chromosome and one copy of the Y chromosome; compared to the typical chromosome makeup where a male has one X chromosome and one Y chromosome. 男性有两个 X 染色体拷贝和一个 Y 染色体拷贝的遗传病；与男性具有一条 X 染色体和一条 Y 染色体的典型染色体组成相比。	克萊恩費爾特氏綜合徵 / 克氏綜合徵 / 先天性睾丸發育不全 (47, XXY)	克兰费尔特综合征 (47, XXY)

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<b>Marker chromosome (n)</b>	<p>A small extra fragment of a chromosome detected when doing a chromosome test like a karyotype. Marker chromosomes can sometimes cause health or development problems, depending on how much and what genetic material is contained within.</p> <p>在进行染色体检查（如核型）时检测到的一小块额外的染色体片段。标记染色体有时会导致健康或发育问题，具体取决于其中所含遗传物质的数量和种类。</p>	標記染色體	标记染色体
<b>Maternal Serum Screening (MSS or maternal serum alphafetoprotein test) (n)</b>	<p>A blood test available to pregnant women that identifies elevated risks for Down syndrome, trisomy 18 and neural tube defects. Usually conducted at in the second trimester, between 14-20 weeks.</p> <p>一种可供孕妇使用的血液检查，可确定唐氏综合症、三染色体 18 症和神经管缺陷的风险升高。通常在孕中期进行，在 14-20 周之间。</p>	孕婦血清篩檢 (MSS 或產婦 血清甲胎蛋白 測試)	母体血清筛查 (MSS 或母体 血清甲胎蛋白 检测)
<b>Microarray (n)</b>	<p>A high-resolution chromosome test that provides more information than a basic karyotype. A microarray measures the amount of chromosome material in a sample and can detect both large changes (e.g., if there is an extra or missing chromosome) as well as small changes (e.g., if there are very small pieces of chromosomes that are extra or missing). It does not look at the visual appearance or arrangement of chromosomes but measures the amount of genetic material.</p> <p>比基本核型提供更多信息的高分辨率染色体检测。微阵列测量样本中染色体物质的数量，并且可以检测到大的变化（例如是否存在额外的或缺失的染色体）以及小的变化（例如是否存在额外的或缺失的非常小的染色体片段）。它不看染色体的视觉外观或排列，而是测量遗传物质的数量。</p>	微陣列技 術 /基因 晶片 /生 物晶片	微阵列
<b>Miscarriage (n)</b>	<p>The spontaneous (not intentional) loss of a pregnancy. See “abortion.” Also called pregnancy loss.</p> <p>自发（非故意）流产。见“abortion”（堕胎）。也称为妊娠丢失。</p>	流產	流产

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<b>Molecule (n)</b>	The smallest unit of a chemical compound that still has the properties of that compound. For example, a molecule of water is made up of two hydrogen atoms and one oxygen atom. Separately, they are just atoms, but when bonded together, they make a water molecule. 化合物的最小单位，仍然具有该化合物的特性。例如，一个水分子由两个氢原子和一个氧原子组成。分开来说，它们只是原子，但当它们结合在一起时，它们会形成一个水分子。	分子	分子
<b>Mosaicism (n)</b>	A condition in which some, but not all, cells in a sample show a genetic difference. 样本中的一些（但不是全部）细胞显示出遗传差异的情况。	鑲嵌現象	镶嵌现象
<b>Multifactorial (adj)</b>	Due to a combination of genetic and non-genetic (environmental, hormonal, etc.) risk factors that act together to determine risk. 由于遗传和非遗传（环境、荷尔蒙等）风险因素，共同决定风险。	多因素的	多因素的
<b>Mutation (n)</b>	A change in a gene, usually deleterious. See “genetic variant.” 基因的改变，通常是有害的。见“遗传变异。” In Spanish, the word is sometimes confused with “mutilation.” 在西班牙语中，这个词有时会与“mutilation”（肢体残缺）混淆。	突變	突变
<b>Non-Invasive Prenatal Testing (NIPT) (n)</b>	A blood test available to pregnant women that identifies elevated risk for certain genetic conditions in the fetus. This test focuses on fragments of DNA from placental cells – which carry the fetus’ genetic make-up – that are found in the mother’s bloodstream. 一种可供孕妇使用的血液检查，可确定胎儿某些遗传疾病的风险升高。这项检查的重点是来自胎盘细胞的DNA片段 – 这些细胞携带胎儿的基因构成 – 在母亲的血液中发现。	非侵入性產前篩檢 (NIPT)	无创产前检测 (NIPT)

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<b>Nuchal thickening (n)</b>	<p>There is a pocket of fluid at the back of the neck of a fetus which can be measured in an ultrasound between 10-14 weeks gestation (called the nuchal translucency). If there is a large amount of fluid at this point, or if later in pregnancy the neck skin itself appears to be thicker, this is associated with a higher risk of chromosome problems and other rare genetic conditions.</p> <p>胎儿颈部后部有一袋液体，可在妊娠 10-14 周之间通过超声波测量（称为颈项透明层）。如果此时有大量液体，或者如果在怀孕后期颈部皮肤本身看起来更厚，这与染色体问题和其他罕见遗传疾病的风险较高有关。</p>	頸背增厚	颈部增厚
<b>Open Neural Tube Defect (ONTD) (n)</b>	<p>A birth defect of the spine, spinal cord, or brain that results from a hole in the spinal column not closing up when appropriate during early fetal development. The two most common types of ONTDs are spina bifida and anencephaly.</p> <p>脊柱、脊髓或大脑的先天缺陷，是由于在胎儿早期发育过程中脊柱上的一个洞没有在适当的时候闭合而导致的。两种最常见的 ONTD 类型是脊柱裂和无脑畸形。</p>	開放性神經管缺陷 (ONTD)	开放性神经管缺陷 (ONTD)
<b>Ova (n)</b>	<p>See “egg.”</p> <p>见 “egg”（卵子）。</p>	卵子	卵子
<b>Ovary (n)</b>	<p>The organ in a woman that stores and releases eggs. There are normally two.</p> <p>女性体内储存和释放卵子的器官。通常有两个。</p>	卵巢	卵巢
<b>Penetrance (n)</b>	<p>The probability that a specific genetic trait will be expressed if a person carries a mutation.</p> <p>“Complete penetrance” means that everyone who carries a particular gene mutation will show the trait related to that altered gene. “Incomplete penetrance” means that only some of the people who have the altered gene will actually show the related trait.</p> <p>表达特定遗传特征的概率，如果一个人携带突变。“完全外显率”意味着每个携带特定基因突变的人都会表现出与该变异基因相关的特征。“不完全外显率”是指只有部分基因发生变异的人会真正表现出相关性状。</p>	外顯率	外显率

Glossary of Prenatal Genetics Terms – Chinese  
产前遗传学术语表

Term/术语	Definition/定义	Traditional/ 繁体中文	Simplified/ 简体中文
<b>Placenta (n)</b>	The organ that develops together with an embryo in a pregnant woman's uterus to nourish the embryo through the umbilical cord. 与胚胎一起在孕妇子宫内发育，通过脐带滋养胚胎的器官。	胎盤	胎盘
<b>Presymptomatic (adj)</b>	Before symptoms appear. 在症状出现之前。	症狀前的	症状前的
<b>Prognosis (n)</b>	The most likely outcome of a disease process. 疾病过程最可能的结果。	病程預測/預後	预后
<b>Protein (n)</b>	A molecule made up of chains of amino acids. Proteins do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs. Genes determine how specific amino acids are put together to form a specific protein. 由氨基酸链组成的分子。蛋白质在细胞中完成大部分工作，是人体组织和器官的结构、功能和调节所必需的。基因决定了特定氨基酸如何组合在一起形成特定蛋白质。	蛋白質	蛋白质
<b>Random (adj)</b>	Happening in an unpredictable way. 以不可预知的方式发生。	隨機的	随机的
<b>Recessive (adj)</b>	A genetic trait in which both copies of a gene need to be altered for the trait to be expressed; recessive traits are typically inherited from both parents, each one contributing one copy of the gene in question. 需要改变基因的两个拷贝才能表达该性状的遗传性状；隐性性状通常从父母双方继承，每人贡献一份相关基因的拷贝。  In prenatal genetics, recessive inheritance refers to conditions in which both copies of a particular gene pair need to be altered to develop a specific disease or condition. 在产前遗传学中，隐性遗传是指特定基因对的两个拷贝都需要改变以发展特定疾病或状况的情况。	隱性的	隐性的
<b>Red flag (n)</b>	A warning sign or a clue. 警告标志或线索。	警告信號	危险信号
<b>Replicate (v)</b>	To copy. 进行拷贝。	複製	复制

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<b>Reproductive history (n)</b>	<p>The experiences a woman has had related to pregnancy and childbirth. A reproductive history usually includes age at the onset of menses (have a monthly period), age at the beginning of sexual intercourse, age at first conception, number of pregnancies, number of live births, number of miscarriages, number of abortions.</p> <p>女性与怀孕和分娩有关的经历。生殖史通常包括月经开始时的年龄（每月一次）、性交开始时的年龄、首次受孕时的年龄、怀孕次数、活产次数、流产次数、堕胎次数。</p>	生產病史	生殖史
<b>Ring chromosome (n)</b>	<p>An abnormal formation of a chromosome in which the ends of two of the chromosome's arms have linked together to form a ring.</p> <p>染色体的异常形成，其中两条染色体臂的末端连接在一起形成一个环。</p>	環狀染色體	环状染色体
<b>Risk(n)</b>	<p>The chance that something will happen; in the case of prenatal genetics, the chance that the child will have a genetic condition.</p> <p>某事件发生的机会；在产前遗传学的情况下，孩子有遗传疾病的机会。</p>	風險	风险
<b>At risk (adj)</b>	<p>Has some possibility of having a genetic condition.</p> <p>有一定的遗传病的可能性。</p>	有風險的	有风险的
<b>At average risk (adj)</b>	<p>Has the same possibility of having a genetic condition as the general population.</p> <p>与一般人群有相同的遗传疾病的可能。</p>	一般風險的	平均风险的
<b>At high risk (adj)</b>	<p>Has a greater possibility of having a genetic condition than the general population.</p> <p>比一般人群有更大的遗传疾病的可能。</p>	高風險的	高风险的
<b>At higher risk than "X" (adj)</b>	<p>Has a greater possibility of having a genetic condition than "X".</p> <p>比 "X" 更有可能患有遗传疾病。</p>	風險高於「X」的	风险高于 "X" 的
<b>Risk factor (n)</b>	<p>A circumstance that increases the risk of having a genetic condition.</p> <p>一种会增加遗传疾病风险的情况。</p>	風險因素	风险因素
<b>Runs in the family (e.g., sickle cell disease runs in the family) (v)</b>	<p>When a certain condition or trait seems to appear in many members of a single family</p> <p>当某个条件或特征似乎出现在一个家庭的许多成员身上时</p>	家族遺傳	家族遗传 (例如镰状细 胞病在家族中 传播)

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<b>Screen (v)</b>	<p>To assess a person's risk for a particular health problem, even if the person has no symptoms to suggest that they have the problem.</p> <p>评估一个人出现特定健康问题的风险，即使该人没有任何症状表明他们有问题。</p>	篩檢	筛查
<b>Screening test (n)</b>	<p>A test that looks to detect a particular health problem, even if a patient has no symptoms of that particular condition.</p> <p>一种旨在检测特定健康问题的试验，即使患者没有该特定疾病的症状。</p> <p>Prenatal screening tests aren't looking for genetic changes. They are designed to look at hormones, chemicals, other things that indicate a fetus might have a higher risk of a certain condition. They are not DNA results, do not give a definite answer about whether someone is affected, and do not provide a firm diagnosis. They are usually reported as a risk number (e.g., 1 in ____ chance), and if this number is above a certain cut-off point, the result will be categorized as 'screen positive' or 'screen negative'.</p> <p>产前筛查检测不是在寻找基因变异。它们旨在查看激素、化学物质和其他表明胎儿可能患某种疾病的风险较高的物质。它们不是DNA结果，没有给出关于某人是否受到影响的明确答案，也没有提供明确的诊断。它们通常被报告为风险数字（例如概率为 ____ 分之 1），如果该数字高于某个分界点，则结果将被归类为“筛查阳性”或“筛查阴性。”</p>	篩檢測試	筛选检测
<b>Semen (n)</b>	<p>A liquid produced by the testes, the prostate gland, the seminal vesicle, and the bulbourethral gland that carries, nourishes and protects sperm cells on their way to fertilizing an egg. Also called seminal fluid.</p> <p>由睾丸、前列腺、精囊和尿道球腺产生的液体，在卵子受精的过程中携带、滋养和保护精子细胞。也称为精液。</p>	精液	精液
<b>Seminal fluid (n)</b>	<p>See "semen."</p> <p>见 "semen"（精液）。</p>	精液	精液

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<b>Sickle cell disease (n)</b>	<p>A genetic disorder caused by a mutation in a gene that helps to make hemoglobin, an important part of red blood cells. In sickle cell disease, red blood cells can become sickle-shaped (instead of round as they should be). These affected blood cells do not function properly, and die early, leaving a shortage of red blood cells, and can block blood flow causing pain.</p> <p>一种由有助于制造血红蛋白的基因突变引起的遗传疾病，血红蛋白是红细胞的重要组成部分。在镰状细胞病中，红细胞会变成镰刀状（而不是应有的圆形）。这些受影响的血细胞功能不正常，过早死亡，导致红细胞不足，并会阻塞血流，导致疼痛。</p>	镰狀細胞疾病	镰状细胞病
<b>Sperm (n)</b>	<p>The reproductive cells of the man. When sperm fertilize a woman's egg, a baby develops.</p> <p>男性的生殖细胞。当精子使女性的卵子受精时，婴儿就会发育。</p>	精子	精子
<b>Sperm donation (n)</b>	<p>The process through which a man allows his sperm to be collected and used to fertilize the eggs of a woman who is not his sexual partner.</p> <p>男人允许他的精子被收集并用于使不是他的性伴侣的女性的卵子受精的过程。</p>	捐精	捐精
<b>Spina bifida (n)</b>	<p>A congenital defect of the spine in which part of the spinal cord is exposed through an opening in the bone structure. This may result in nerve damage and some degree of paralysis in the legs.</p> <p>脊柱的一种先天性缺陷，其中部分脊髓通过骨骼结构中的开口暴露出来。这可能会导致神经损伤和腿部某种程度的麻痹。</p>	脊柱裂	脊柱裂
<b>Statistically significant (adj)</b>	<p>A measurement of whether the findings of research are meaningful. Refers to the likelihood that a relationship between two factors is linked by something other than chance.</p> <p>衡量研究结果是否有意义。指两个因素之间的关系由非偶然因素联系起来的可能性。</p>	有統計意義的	有统计学意义的
<b>Teratogen (n)</b>	<p>Something that has the potential to disturb the normal development of an embryo or fetus.</p> <p>有可能干扰胚胎或胎儿正常发育的物质。</p>	畸胎原	致畸原
<b>Termination of pregnancy (n)</b>	<p>See "abortion."</p> <p>见“abortion”（堕胎）。</p>	終止妊娠	终止妊娠

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<b>Test result (n)</b>	A negative result on a genetic test means that the laboratory did not find the specific genetic change that the test was designed to identify. 基因检测的阴性结果意味着实验室没有发现检测旨在识别的特定基因变化。	測試結果 陰性的	检测结果 阴性的
<b>True negative (n)</b>	When a patient is being tested to see if he or she has a specific genetic change that is present in other family members, a negative result can be considered a “true negative.” This means that instead of having an increased risk for cancer like other family members, they have the same risk as everyone else in the general population. 当对患者进行检测以查看他或她是否具有其他家庭成员中存在的特定基因变化时，阴性结果可被视为“真阴性。”这意味着他们不会像其他家庭成员那样增加患癌症的风险，而是与普通人群中的其他人具有相同的风险。	真陰性	真阴性
<b>Positive (adj)</b>	A positive result on a genetic test means that the laboratory did find a specific genetic change that is associated with a hereditary cancer syndrome (a deleterious mutation). In cancer genetics, that means that the patient has an increased risk of getting the cancer(s) associated with the gene. 基因检测的阳性结果意味着实验室确实发现了与遗传性癌症综合征（有害突变）相关的特定基因变化。在癌症遗传学中，这意味着患者患上与该基因相关的癌症的风险增加。 If the laboratory finds a genetic change for which currently there is not enough information to know if this change is problematic or not, it reports a “variant of uncertain significance.” Basically, this means that the laboratory found a genetic change, but they don’t know what it means. All of these variants will eventually be reclassified as either a “positive” or “negative” result. Most become “negative” and are thought to represent natural variation between individuals. 如果实验室发现目前没有足够信息说明这种变化是否有问题的基因变化，它会报告“具有不确定意义的变异。”基本上，这意味着实验室发现了基因变化，但他们不知道这意味着什么。所有这些变异最终将被重新分类为“阳性”或“阴性”结果。大多数变为“阴性”，并被认为代表了个人之间的自然差异。	陽性的	阳性的
<b>Variant of uncertain significance (n)</b>	Everyone has some degree of commonly occurring genetic changes that are not associated with cancer. If the test finds this sort of change, it reports a “polymorphism.”	意義不明確的 變異	意义不明确的 变异
<b>Polymorphism (n)</b>		多態現象 / 多態性 / 多型性	多态性

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	每个人都都有一定程度的与癌症无关的常见基因变化。如果检测发现这种变化，它会报告“多态性。”		
<b>Testes (n)</b>	The organs in a man that create and release sperm. Also called “testicles.” Singular testis. 男人体内产生和释放精子的器官。也称为“睾丸。”单睾丸。	睾丸	睾丸
<b>Toxic exposure (n)</b>	Contact with something that is harmful or poisonous. 接触有害或有毒的物质。	毒性暴露 / 接触有毒物	毒性暴露
<b>Trait (n)</b>	A characteristic of a person. 一个人的特征。 In genetics, traits are aspects of a person defined or influenced by their genetic code. E.g., Eye color, blood type, risk for certain diseases. 在遗传学中，特征是受遗传密码定义或影响的人的某些方面。例如眼睛颜色、血型、某些疾病的风险。	特徵	特征
<b>Translocation (n)</b>	Translocations are a type of chromosome rearrangement. They can be ‘balanced’, meaning that two chromosomes have pieces that have ‘swapped’ with each other, but there is still the correct amount of chromosome material. Balanced translocations do not usually cause medical or development problems. They can also be ‘unbalanced’, where pieces of chromosomes have traded places, but there is missing or extra chromosome material. Unbalanced translocations will often cause medical and development problems. 易位是染色体重排的一种。它们可以是“平衡的”，这意味着两条染色体的片段相互“交换”，但仍然有正确数量的染色体物质。平衡易位通常不会导致医疗或发育问题。它们也可能是“不平衡的”，即染色体片段交换了位置，但出现缺少或多余的染色体材料。不平衡易位通常会导致医疗和发育问题。	移位/轉移	易位

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<b>Typo (n)</b>	<p>A mistyped word; shortened slang for “typographical error.”  一个打错的字；“印刷错误”的缩写俚语。  In genetic counseling “a typo” is commonly used as an analogy to refer to mutations or variants in a gene.  在遗传咨询中，“错字”通常用作类比来指代基因中的突变或变异。</p>	串錯字	错字
<b>Ultrasound (n)</b>	<p>An imaging method that uses high frequency sound waves to create a picture of something inside the body, such as a fetus or baby  一种成像方法，其使用高频声波来创建体内某物质的图像，例如胎儿或婴儿。</p>	超聲波	超声波
<b>Uterus (n)</b>	<p>The organ in a woman’s lower abdomen in which a fertilized egg develops into an embryo, then a fetus, then a baby. Also called “womb.”  女性下腹部的器官，受精卵在其中发育成胚胎，然后是胎儿，然后是婴儿。也称为“子宫”。</p>	子宫	子宫
<b>Vagina (n)</b>	<p>The tube-like part of the female reproductive system that extends from the cervix to the outside of the body.  女性生殖系统的管状部分，从子宫颈延伸到体外。</p>	陰道	阴道
<b>Variant (n)</b>	<p>A version of something that differs from the norm. So, a genetic variant is a change to the usual genetic sequence.  与规范不同的物质的版本。因此，遗传变异是对通常基因序列的改变。</p>	變異	变异
<b>Vas deferens (n)</b>	<p>The tubes that lead from the testes to the urethra, through which sperm is ejaculated.  从睾丸通向尿道的管子，精子通过这些管子射出。</p>	輸精管	输精管
<b>X-linked (adj)</b>	<p>A trait that is influenced or determined by a gene on the “X” chromosome.  受“X”染色体上的基因影响或决定的性状。</p>	X 染色體串聯的	X 连锁的

## Family Relationships

### 家庭关系

For genetic counselors, it is very important to know if a relative is a blood relative or a relative by marriage.

对于遗传咨询师来说，了解亲属是血缘亲属还是婚姻亲属非常重要。

Term 术语	Definition 定义	Traditional 繁体中文	Simplified 简体中文
<b>Parent</b>	Your mother or father. 你的母亲或父亲。	父母	父母
<b>Mother</b>	For genetic purposes, the woman whose egg was fertilized and grew to be you. 出于遗传目的，卵子受精并成长为你的女性。	母親	母亲
<b>Mother-in-law</b>	Your husband or wife's mother. 你丈夫或妻子的母亲。	岳母/婆婆	岳母
<b>Stepmother</b>	Your father's wife who is not your biological mother. 你父亲的妻子，不是你的亲生母亲。	繼母	继母
<b>Adoptive mother</b>	A woman who is not your biological mother but who accepted legal responsibility for and raised you. 一个不是你亲生母亲但为你承担法律责任并抚养你长大的女性。	養母	养母
<b>Godmother</b>	A woman chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church. 一个被你父母选为你生命中精神导师的女性，通过天主教堂的仪式命名。	教母 契母/ 契媽 (Formal/informal acceptance of kinship in a non-Christian context)	教母

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<b>Father</b>	For genetic purposes, the man whose sperm fertilized the egg that grew to be you. 出于遗传目的，精子使卵子受精，然后成长为你的男性。	父親	父亲
<b>Father-in-law</b>	Your husband or wife's father. 你丈夫或妻子的父亲。	岳父/公公 老爺 / 家公	岳父
<b>Stepfather</b>	Your mother's husband who is not your biological father. 你母亲的丈夫，不是你的亲生父亲。	繼父	继父
<b>Adoptive father</b>	A man who is not your biological father but who accepted legal responsibility for and raised you. 一个不是你亲生父亲但为你承担法律责任并抚养你长大的男性。	養父	养父
<b>Godfather</b>	A man chosen by your parents to be your spiritual guide in life, named as such through a ceremony in the Catholic church. 一个被你的父母选为你生命中精神导师的男性，通过天主教堂的仪式命名。	契父/ 契爺 (formal/informal acceptance of kinship in a non-Christian context)	教父
<b>Aunt</b>	Your mother or father's sister or sister-in-law. 你母亲或父亲的姐妹或嫂子/弟妹。	阿姨 (younger aunt) 姨媽 (older aunt)	姨妈/舅妈/姑妈/ 伯母/叔母
<b>Maternal aunt</b>	Your mother's sister or sister-in-law. 你母亲的姐妹或嫂子/弟妹。	舅母 / 舅媽 (wife of mother's older brother)	姨妈/舅妈
<b>Paternal aunt</b>	Your father's sister or sister-in-law. 你父亲的姐妹或嫂子/弟妹。	姑姑 / 姑媽 (father's older sister/ father's younger sister) 嬸嬸 (wife of father's younger brother) 伯母 (wife of father's older brother)	姑妈/伯母/叔母

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<b>Uncle</b>	Your mother or father's brother or brother-in-law. 你母亲或父亲的兄弟或姐夫/妹夫。	小舅 (younger uncle) 大舅 (older uncle)	舅舅/姨夫/伯父/ 叔叔/姑父
<b>Maternal uncle</b>	Your mother's brother or brother-in-law. 你母亲的兄弟或姐夫/妹夫。	姨丈 (husband of mother's sister)	舅舅/姨夫
	Your father's brother or brother-in-law. 你父亲的兄弟或姐夫/妹夫。	叔叔/叔父(father's younger brother) 伯父 (father's older brother) 姑丈/姑爹(husband of father's sister)	伯父/叔叔/姑父
<b>Cousin</b>	Usually understood to be a first cousin. 通常被理解为堂/表兄弟姐妹。	堂(表)兄弟姊妹	堂/表兄弟姐妹
	Your aunt or uncle's child. 你的姨妈/舅妈/姑妈/伯母/叔母或舅舅/姨夫/伯父/叔叔/姑父的孩子。	堂兄弟姊妹(father's side)	堂/表侄子/侄女
	Your aunt or uncle's grandchild or your first cousin's child 你的姨妈/舅妈/姑妈/伯母/叔母或舅舅/姨夫/伯父/叔叔/姑父的孙子或你的堂/表兄弟姐妹的孩子。	表兄弟姊妹 (mother's side)	远房堂/表侄子/ 侄女
	The children of first cousins are second cousins to each other. 堂/表兄弟姐妹的孩子是彼此的远房堂/表兄弟姐妹。	远房堂兄弟姊妹 (father's side)  远房表兄 弟姊妹 (mother's side)	
<b>Grandparent</b>	Your parent's father or mother. 你的父母的父亲或母亲。	祖父母	外祖父母/ 祖父母
<b>Grandmother</b>	Your mother or father's mother.	祖母	外祖母/祖母
<b>Maternal grandmother</b>	Your mother's mother. 你的母亲的母亲。	阿婆	外祖母
<b>Paternal grandmother</b>	Your father's mother. 你的父亲的母亲。	阿嬷	祖母

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<b>Maternal grandfather</b>	Your mother's father. 你的母亲的父亲。	外公/阿公	外祖父
<b>Paternal grandfather</b>	Your father's father. 你的父亲的父亲。	阿爺/爺爺	祖父
<b>Great aunt</b>	Your mother or father's aunt. 你的母亲或父亲的姨妈/舅妈/姑妈/伯母/叔母。	姑婆 / 伯婆 / 姮婆 / 姨婆 / 叔婆	姑/姨祖母
<b>Maternal great aunt</b>	Your mother's aunt. 你的母亲的姨妈/舅妈/姑妈/伯母/叔母。	姑婆 / 伯婆 / 姮婆 / 姨婆 / 叔婆	姨祖母
<b>Paternal great aunt</b>	Your father's aunt. 你的父亲的姨妈/舅妈/姑妈/伯母/叔母。	姑婆 / 伯婆 / 姮婆 / 姨婆 / 叔婆	姑祖母
<b>Great uncle</b>	Your father or mother's uncle. 你的父亲或母亲的舅舅/姨夫/伯父/叔叔/姑父。	舅公 / 姑公 / 姨公	伯/舅祖父
<b>Maternal great uncle</b>	Your mother's uncle. 你的母亲的舅舅/姨夫/伯父/叔叔/姑父。	伯祖(父)(older brother) 叔公(younger brother)	舅祖父
<b>Paternal great uncle</b>	Your father's uncle. 你的父亲的舅舅/姨夫/伯父/叔叔/姑父。	叔公(younger brother)	伯祖父
<b>Great grandparents</b>	The parents of any of your grandparents. 你的任何祖父母的父母。	曾祖父母	曾祖父母
<b>Great grandmother</b>	The mother of any of your grandparents. 你的任何祖父母的母亲。	曾祖母	曾祖母
<b>Great grandfather</b>	The father of any of your grandparents. 你的任何祖父母的父亲。	曾祖父	曾祖父

Glossary of Prenatal Genetics Terms – Chinese  
产前遗传学术语表

Term 术语	Definition 定义	Traditional 繁体中文	Simplified 简体中文
<b>siblings</b>	The children of your father and mother. 你的父亲和母亲的孩子。	兄弟姊妹	兄弟姐妹
<b>Half siblings</b>	Siblings (brothers and sisters) who have either the same mother and different fathers, or the same father but different mothers. Half siblings share some genetic similarity with you. 同母异父或同父异母的胞兄弟姐妹（兄弟姐妹）。胞兄弟姐妹与你有一些遗传相似性。	同父異母或同母異父的兄弟姊妹	胞兄弟姐妹
<b>Stepsiblings</b>	The children of your stepmother but not your father; or the children of your stepfather but not your mother. Stepsiblings do not share any genetic similarity with you. 你继母的孩子，但不是你父亲的孩子；或者你继父的孩子，但不是你母亲的孩子。继兄弟姐妹与你没有任何遗传相似性。	繼兄弟姊妹	继兄弟姐妹
<b>Twins</b>	Two siblings born at the same time. 两个兄弟姐妹同时出生。	雙胞胎	双胞胎
<b>Identical twins</b>	Twins who developed from the same egg and sperm, meaning that they are genetically identical. Also called monozygotic twins. 从同一个卵子和精子发育而来的双胞胎，这意味着他们在基因上是相同的。也称为同卵双胞胎。	同卵雙胞胎	同卵双胞胎
<b>Fraternal twins</b>	Twins who developed from different eggs and sperm, meaning that they are genetically different and have the same number of shared genes as any other sibling. Also called dizygotic twins. 从不同的卵子和精子发育而来的双胞胎，这意味着他们在基因上是不同的，并且与任何其他兄弟姐妹具有相同数量的共享基因。也称为异卵双胞胎。	異卵雙胞胎	异卵双胞胎
<b>Triplets</b>	Three siblings born at the same time. 三个兄弟姐妹同时出生。	三胞胎	三胞胎

Term 术语	Definition 定义	Traditional 繁体中文	Simplified 简体中文
<b>Sister</b>	A sibling who is a girl. 是女孩的兄弟姐妹。	姊妹	姐妹
<b>Sister-in-law</b>	Your brother's wife or your husband's sister. 你兄弟的妻子或你丈夫的姐妹。	嫂嫂/嫂子 (older brother's wife) 弟媳(younger brother's wife)	嫂子/弟妹/大姑子/小姑子
<b>Half sister</b>	A girl who is either the child of your father with a different mother, or the child of your mother with a different father. 一个女孩，要么是你父亲和另一个母亲的孩子，要么是你母亲和另一个父亲的孩子。	同父異母姊妹 (same father) 同母異父姊妹 (same mother)	胞姐妹
<b>Stepsister</b>	The daughter of your stepmother or stepfather and therefore not biologically related to you. 你的继母或继父的女儿，因此与你没有生物学上的关系。	繼姊妹	继姐妹
<b>Brother</b>	A sibling who is a boy. 是男孩的兄弟姐妹。	兄弟	兄弟
<b>Brother-in-law</b>	Your sister's husband or your wife's brother. 你姐姐的丈夫或你妻子的兄弟。	姊夫(older sister's) 妹夫 (younger sister's)	姐夫/妹夫/妻兄/妻弟
<b>Half brother</b>	A boy who is either the child of your father with a different mother, or the child of your mother with a different father. 一个男孩，要么是你父亲和另一个母亲的孩子，要么是你母亲和另一个父亲的孩子。	同父異母兄弟 (same father) 同母異父兄弟 (same mother)	胞兄弟
<b>Stepbrother</b>	The son of your stepmother or stepfather and therefore not biologically related to you. 你的继母或继父的儿子，因此与你没有生物学上的关系。	繼兄弟	继兄弟
<b>Niece</b>	Your sibling's daughter. 你兄弟姐妹的女儿。	姪女(brother's daughter) 外甥女(sister's daughter)	侄女/外甥女
<b>Nephew</b>	Your sibling's son. 你兄弟姐妹的儿子。	姪子(brother's son) 外甥(sister's son)	侄子/外甥
<b>Spouse</b>	Your husband or wife. 你的丈夫或妻子。	配偶	配偶

Term 术语	Definition 定义	Traditional 繁体中文	Simplified 简体中文
<b>Wife</b>	The woman to whom you are married. 与你结婚的女人。	妻子	妻子
<b>Husband</b>	The man to whom you are married. 与你结婚的男人。	丈夫	丈夫
<b>Children</b>	Genetically speaking, the people who are produced from your egg or sperm. 从基因上讲，从你的卵子或精子中产生的 人。	子女	孩子
<b>Daughter</b>	Your child who is a girl. 是女孩的孩子。	女兒	女儿
<b>Daughter-in-law</b>	Your son's wife. 你儿子的妻子。	媳婦	儿媳
<b>Stepdaughter</b>	Your spouse's daughter who is not your biological child. 你配偶的女儿，不是你的亲生孩子。	繼女	继女
<b>Adoptive daughter</b>	A girl for whom you have accepted legal responsibility and raised even though she is not your biological child. 一个女孩，即使她不是你的亲生孩子，你也已为其承担法律责任并抚养其长大。	養女	养女
<b>Goddaughter</b>	A girl for whom you have accepted spiritual responsibility within a ceremony of the Catholic church; she may or may not be biologically related to you. 一个女孩，你在天主教会的仪式中为她承担精神责任；她可能与你有生物学上的关系，也可能没有。	教女 契女 (formal/informal acceptance of kinship in a non-Christian context)	教女

Term 术语	Definition 定义	Traditional 繁体中文	Simplified 简体中文
<b>Son</b>	Your child who is a boy. 是男孩的孩子。	兒子	儿子
<b>Son-in-law</b>	Your daughter's husband. 你女儿的丈夫。	女婿	女婿
<b>Stepson</b>	Your spouse's son who is not your biological child.	繼子	继子
<b>Adoptive son</b>	你配偶的儿子，不是你的亲生孩子。 A boy for whom you have accepted legal responsibility and raised even though he is not your biological child.	養子	养子
<b>Godson</b>	一个男孩，即使他不是你的亲生孩子，你也已为其承担法律责任并抚养其长大。 A boy for whom you have accepted spiritual responsibility within a ceremony of the Catholic church. He may or may not be biologically related to you. 一个男孩，你在天主教会的仪式中为她承担精神责任。他可能与你有生物学上的关系，也可能没有。	教子 契子/契仔 (Formal/informal acceptance of kinship in a non-Christian context)	教子
<b>Ancestor</b>	A person from whom you are descended, usually more remote than a grandparent. 你的祖辈，通常比祖父母更遥远。	祖先	祖先
<b>Fiancé(e)</b>	The person whom you have promised to marry. 你答应与其结婚的人。 Man: fiancé 男：未婚夫 Woman: fiancée 女：未婚妻	未婚夫（妻）	未婚夫/妻
<b>Domestic partner</b>	The person with whom you are living and have an intimate relationship, but to whom you are not married. 与你同住并有亲密关系但未结婚的人。	同居伴侣	同居伴侣
<b>Divorced</b>	Having ended a marriage. 结束了一段婚姻。	離婚	离婚
<b>Engaged</b>	Having promised to marry someone. 答应嫁给别人。	訂婚	订婚
<b>Widow</b>	A woman whose husband has died. 丈夫去世的女人。	寡婦 /遺孀	寡妇
<b>Widower</b>	A man whose wife has died. 妻子去世的男人。	鳏夫	鳏夫

## Analogies

Genetic counselors often use analogies to explain complex ideas in genetics. An analogy explains a new concept by comparing it to something more common. A clue that an analogy is being used are the words “It’s as if” and “It’s like . . . .”

Examples:

“Genes are like an instruction book or an instruction manual. And in a book, there is a certain sequence to a story.”

“It's as if you were reading through a long book and looking for one typo, for a letter that is mistaken.”

“So, a mutation is like an unexpected change in the order of the letters.”

“A gene is like a recipe, and mutations are like changes in that recipe.”

“It's like chapters in a book, and like misspellings.”

“Our cells are like libraries and our chromosomes are like books.”

“Like a flip of a coin.” (to explain that there is a 50% chance that a mutation will be passed on every time the patient has a child).

“Inside every cell are genes, which are like instruction books for our bodies; they tell our bodies how to grow and develop.”

“It is kind of like you reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for.”

Some analogies may not work with certain patients, due to linguistic or experiential differences.

Analogies that focus on genes as “letters in a book” may make no sense to speakers of character-based written languages such as Chinese, Japanese, and Korean.

“It's as if you were reading through a long book and looking for one typo, for a letter that is mistaken.”

“So a mutation is like an unexpected change in the order of the letters.”

“It's like chapters in a book, and like misspellings.”

“It is kind of like if you are reading a book and you notice that a word is spelled wrong; that is what this genetic test looks for.”

Analogies that focus on genes as a recipe may not work for patients who do not cook with written recipes.

“A gene is like a recipe, and mutations are like changes in that recipe.”

Analogies that discuss random chance in terms of a coin toss may not work for patients from cultures where people don't flip coins to make decisions.

“Like a flip of a coin” (to explain that there is a 50% chance that a mutation will be passed on every time the patient has a child).

## 类比

遗传咨询师经常使用类比来解释遗传学中的复杂想法。类比通过将一个新概念与更常见的事物进行比较来解释它。使用类比的线索是“这好像”和“这就像...”这两句话。

举例：

“基因就像一本说明书或说明手册。在一本书中，一个故事总有一定的发展顺序。”

“这好像你正在阅读一本长书，寻找一个错字，其中的一个字母写错了。”

“所以，突变就像字母顺序的意外变化。”

“基因就像一个食谱，而突变就像那个食谱的变化。”

“这就像一本书的章节，就像拼写错误。”

“我们的细胞就像图书馆，我们的染色体就像图书。”

“就像掷硬币一样。”（解释每次患者生孩子时有 50% 的机会遗传突变）。

“每个细胞里面都有基因，它就像我们身体的指导书；它们告诉我们的身体如何成长和发育。”

“这有点像你正在读一本书，你发现一个单词写错了；这就是这项基因检测所要寻找的。”

由于语言或经验差异，某些类比可能不适用于某些患者。

将基因比作“书中的字母”的类比对于使用基于字符的书面语言（如中文、日文和韩文）的人来说可能没有意义。

“这好像你正在阅读一本长书，寻找一个错字，其中的一个字母写错了。”

“所以，突变就像字母顺序的意外变化。”

“这就像一本书的章节，就像拼写错误。”

“这有点像你正在读一本书，你发现一个单词写错了；这就是这项基因检测所要寻找的。”

将基因比作食谱的类比可能不适用于不使用书面食谱烹饪的患者。

“基因就像一个食谱，而突变就像那个食谱的变化。”

用掷硬币来讨论随机机会的类比可能不适用于来自人们不掷硬币做出决定的文化的患者。

“就像掷硬币一样”（解释每次患者生孩子时有 50% 的机会遗传突变）。

Glossary of Prenatal Genetics Terms – Chinese  
产前遗传学术语表

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