

Date of Birth
09/25/1950Sex
FemalePhysician
Li-Yuan BaiInstitution
China Medical University Hospital
12357270**TEMPUS | xF**

105 gene liquid biopsy

cfDNA specimen:
Peripheral Blood
Collected 8/21/2023
Received 8/24/2023**GENOMIC VARIANTS****Biologically Relevant**

Biologically Relevant	Variant Allele Fraction
<div style="display: flex; align-items: center;"> <div style="border: 1px solid black; border-radius: 15px; padding: 2px 10px; margin-right: 10px;"> ↔ NF1 </div> <div> <p>c.7869+1G>A Splice region variant - LOF</p> </div> </div>	<p>0.4% ▬</p>

Median Variant Allele Fraction

0.4%

IMMUNOTHERAPY MARKERS**Microsatellite Instability Status**



MSI-High not detected

TREATMENT IMPLICATIONS

No reportable treatment options found.

CLINICAL TRIALS



PF-07284892 in Participants With Advanced Solid Tumors (NCT04800822)	Phase I Grand Rapids, MI - 130 mi ✓ NF1 c.7869+1G>A mutation
Tovorafenib (DAY101) Monotherapy or in Combination With Other Therapies for Patients With Melanoma and Other Solid Tumors (NCT04985604)	Phase I/II Indianapolis, IN - 159 mi ✓ NF1 c.7869+1G>A mutation
A Basket Trial of an ERK1/2 Inhibitor (LY3214996) in Combination With Abemaciclib. (NCT04534283)	Phase II Indianapolis, IN - 164 mi ✓ NF1 c.7869+1G>A mutation

Gene	Mutation effect	Variant allele fraction
ARID1A	c.547G>A p.A183T Missense variant NM_006015	50.9% 
NF1	c.1007G>T p.W336L Missense variant NM_001042492	0.9% 

LOW COVERAGE REGIONS

ERRFI1 JAK1 KMT2A MSH3 SPOP TERT

VARIANT DETAILS - BIOLOGICALLY RELEVANT

 **NF1** c.7869+1G>A NM_001042492 Splice region variant - LOF VAF: 0.4% 

NF1 is a tumor suppressor that plays a role in cellular growth and differentiation through the regulation of the Ras protein. Loss of function mutations and copy number loss of NF1 are associated with cancer progression.

Assay Description

The Tempus xF assay is a next-generation sequencing (NGS) cell-free DNA liquid biopsy tumor profiling assay for identifying genomic alterations derived from solid tumors but circulating in the blood. The 105 gene panel includes single nucleotide variants (SNVs), insertions and deletions (indels), copy number variants (CNVs) and chromosomal rearrangements (translocations) detected by hybrid capture NGS using custom designed IDT probes. The assay typically uses 30 to 50 ng of input DNA, and at 30 ng of input material, the technical sensitivity is >99% for SNVs and CNV amplifications at ≥0.5% variant allele fraction (VAF), and >98% for indels and >97% for translocations at >0.5% VAF. The assay spans clinically relevant coding exons for 35 genes and covers recurrent hotspot mutations in 70 genes. Insertions and deletions will be reported down to the lower limit of detection (LLOD) in clinically relevant regions in 97 genes (list available upon request). BRCA1 and BRCA2 copy number losses are reported when detected. At the discretion of the attending pathologists, the assay may be run at 10 to <30 ng of input DNA, but in such a case, the report will indicate reduced sensitivity and consideration should be given to additional testing. Please see the [Tempus website](#) for a complete gene list and performance specifications.

Potentially Actionable alterations are protein-altering variants with an associated therapy, diagnostic, and/or prognostic indication, based on evidence from clinical guidelines and medical literature. **Biologically Relevant** variants have functional significance or an association with the disease state in the medical literature, but do not have relevant therapeutic, prognostic or diagnostic evidence in the Tempus knowledge database. **Variants of Unknown Significance (VUSs)** exhibit an unclear effect on function and/or do not have sufficient evidence to determine their pathogenicity. **Benign variants** are not reported. **Low Coverage Regions** are included when mean coverage over any region(s) of a gene falls below a threshold of 1000x. The absence of alterations in genes with low coverage should be interpreted carefully in the context of the patient's diagnosis with consideration for retesting. Variants are identified through aligning the patient's DNA sequence to the human genome reference sequence version hg19 (GRCh37). The clinical summary shows actionable and biologically relevant variants. Because sequencing is performed without a matched normal sample, it is not possible to distinguish whether reported variants are germline or somatic.

Microsatellite instability (MSI) refers to hypermutability caused by genetic or acquired defects in the DNA mismatch repair pathway. **MSI-high (MSI-H)** tumors have changes in microsatellite repeat lengths due to defective DNA mismatch repair activity. MSI-H status is reported when detected. If MSI status will affect clinical management, immunohistochemical staining for DNA mismatch repair proteins, or another method of ascertaining MSI status, is recommended.

xF provides insights into the clinically relevant biomarkers incorporated in OncoKB, NCCN and other oncology guidelines for:

Bladder cancer: FGFR2, FGFR3

Breast cancer: BRCA1, BRCA2, ERBB2 (HER2), ESR1, PIK3CA

Cholangiocarcinoma: FGFR2, IDH1

Colorectal cancer: BRAF, ERBB2 (HER2), KRAS, NRAS

Assay Description (continued)**Gastroesophageal adenocarcinoma:** ERBB2 (HER2)**Gastrointestinal stromal tumor:** KIT, PDGFRA**Melanoma:** BRAF, KIT, NRAS**Non-small cell lung cancer:** ALK, BRAF, EGFR, ERBB2 (HER2), KRAS, MET, RET, ROS1**Complete Gene List**

A-C

AKT1, AKT2, ALK, APC, AR, ARAF, ARID1A, ATM, ATR, B2M, BAP1, BRAF, BRCA1, BRCA2, BTK, CCND1, CCND2, CCND3, CCNE1, CD274 (PD-L1), CDH1, CDK4, CDK6, CDKN2A, CTNNA1

D-F

DDR2, DPYD, EGFR, ERBB2 (HER2), ERFF1, ESR1, EZH2, FBXW7, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FOXL2

G-M

GATA3, GNA11, GNAQ, GNAS, HNF1A, HRAS, IDH1, IDH2, JAK1, JAK2, JAK3, KDR, KEAP1, KIT, KMT2A, KRAS, MAP2K1, MAP2K2, MAPK1, MET, MLH1, MPL, MSH2, MSH3, MSH6, MTOR, MYC, MYCN

N-R

NF1, NF2, NFE2L2, NOTCH1, NPM1, NRAS, NTRK1, PALB2, PBRM1, PDCD1LG2, PDGFRA, PDGFRB, PIK3CA, PIK3R1, PMS2, PTCH1, PTEN, PTPN11, RAD51C, RAF1, RB1, RET, RHEB, RHOA, RIT1, RNF43, ROS1

S-Z

SDHA, SMAD4, SMO, SPOP, STK11, TERT, TP53, TSC1, TSC2, UGT1A1, VHL

Gene Rearrangements

ALK, BRAF, FGFR2, FGFR3, NTRK1, RET, ROS1

Copy Number Gains

CCNE1, CD274 (PD-L1), EGFR, ERBB2 (HER2), MET, MYC

Copy Number Losses

BRCA1 and BRCA2

Tempus Disclaimer

The analysis of nucleic acids by next-generation sequencing (NGS) can be affected by multiple factors including DNA quality, hemolysis of the peripheral blood sample, and low amounts of circulating cell-free DNA limiting sensitivity. Tempus may report findings below our sensitivity threshold due to reduced sample quality and/or quantity. For samples flagged as falling below this threshold, Tempus advises resequencing in order to provide more accurate results. Additionally, the chance of detecting genetic alterations may be reduced in regions of the genome which are structurally difficult to sequence, in homologous genes, or due to sequencing errors.

Genetic alterations are defined as clinically significant based on peer-reviewed published literature and other authoritative sources such as NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®). These references are not comprehensive, therefore clinically unknown findings may occur.

These test results and information contained within the report are current as of the report date. Tempus will not update reports or send notification regarding reclassification of genomic alterations. This test was developed and its performance characteristics determined by Tempus Labs, Inc. It has not been cleared or approved by the US Food and Drug Administration. The laboratory is CLIA certified to perform high-complexity testing. Any decisions related to patient care and treatment choices should be based on the independent judgment of the treating physician and should take into account all information related to the patient, including without limitation, the patient and family history, direct physical examination and other tests. Tempus is not liable for medical judgment with regards to diagnosis, prognosis or treatment in connection with the test results.

Note that certain tumor type-, sample- or variant-related characteristics, such as low cell-free DNA concentration, may result in reduced analytic sensitivity of the xF test for detection of alterations in the covered genes, including the above mentioned guideline-recommended genes.

Dostarlimab is indicated for the treatment of patients with mismatch repair deficient (dMMR) endometrial cancer or solid tumors. Although patients with MSI-H cancers are likely dMMR, confirmatory testing is recommended.

Tempus Disclaimer (continued)

Dates and times are represented in the coordinated universal time zone (UTC) unless otherwise specified. However, dates that are provided to Tempus without a timestamp (e.g., sample collection date) are listed as provided.

This assay cannot distinguish whether cell-free variants detected in plasma are derived from a patient's solid tumor or from clonal blood cells. Variants associated with clonal hematopoietic processes may be detected in a high percentage of older individuals and are especially common in the following genes:

ATM, BRAF, BRCA1, BRCA2, EZH2, FLT3, GNAS, IDH1, IDH2, JAK2, KIT, KRAS, NOTCH1, NPM1, NRAS, PALB2, PTPN11, RAD51C, RHOA, TP53

Correlation with tumor and/or blood sequencing results may be helpful in clarifying the origin of variants in some cases.

1. Genovese G, Kahler AK, Handsaker RE, et al. Clonal hematopoiesis and blood-cancer risk inferred from blood DNA sequence. *N Engl J Med.* 2014;371:2477-2487.
2. Jaiswal S, Fontanillas P, Flannick J, et al. Age-related clonal hematopoiesis associated with adverse outcomes. *N Engl J Med.* 2014;371:2488-2498.
3. Chan HT, Chin YM, Nakamura Y, et al. Clonal Hematopoiesis in Liquid Biopsy: From Biological Noise to Valuable Clinical Implications. *Cancers* 2020, 12, 2277. <https://doi.org/10.3390/cancers12082277>

Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) Acute Lymphoblastic Leukemia Version: 1.2022 Acute Myeloid Leukemia Version: 3.2023 Ampullary Adenocarcinoma Version: 1.2023 Anal Carcinoma Version: 2.2023 Basal Cell Skin Cancer Version: 1.2023 B-Cell Lymphomas Version: 2.2023 Biliary Tract Cancers Version: 1.2023 Bladder Cancer Version: 2.2023 Bone Cancer Version: 3.2023 Breast Cancer Version: 4.2023 Central Nervous System Cancers Version: 1.2023 Cervical Cancer Version: 1.2023 Chronic Lymphocytic Leukemia/Small Lymphocytic Lymphoma Version: 2.2023 Chronic Myeloid Leukemia Version: 2.2023 Colon Cancer Version: 2.2023 Dermatofibrosarcoma Protuberans Version: 1.2023 Esophageal and Esophagogastric Junction Cancers Version: 2.2023 Gastric Cancer Version: 1.2023 Gastrointestinal Stromal Tumors Version: 1.2023 Gestational Trophoblastic Neoplasia Version: 1.2023 Hairy Cell Leukemia Version: 1.2023 Head and Neck Cancers Version: 1.2023 Hepatobiliary Cancers Version: 1.2023 Hepatocellular Carcinoma Version: 1.2023 Histiocytic Neoplasms Version: 1.2022 Hodgkin Lymphoma Version: 2.2023 Kaposi Sarcoma Version: 1.2023 Kidney Cancer Version: 4.2023 Melanoma: Cutaneous Version: 2.2023 Melanoma: Uveal Version: 1.2023 Merkel Cell Carcinoma Version: 1.2023 Mesothelioma: Peritoneal Version: 1.2023 Mesothelioma: Pleural Version: 1.2023 Multiple Myeloma Version: 3.2023 Myelodysplastic Syndromes Version: 1.2023 Myeloid/Lymphoid Neoplasms with Eosinophilia and Tyrosine Kinase Fusion Genes Version: 2.2022 Myeloproliferative Neoplasms Version: 3.2022 Neuroendocrine and Adrenal Tumors Version: 2.2022 Non-Small Cell Lung Cancer Version: 3.2023 Occult Primary Version: 3.2023 Ovarian Cancer/Fallopian Tube Cancer/Primary Peritoneal Cancer Version: 1.2023 Pancreatic Adenocarcinoma Version: 1.2023 Pediatric Acute Lymphoblastic Leukemia Version: 2.2023 Pediatric Aggressive Mature B-Cell Lymphomas Version: 1.2023 Pediatric Central Nervous System Cancers Version: 2.2023 Pediatric Hodgkin Lymphoma Version: 2.2023 Penile Cancer Version: 1.2023 Primary Cutaneous Lymphomas Version: 1.2023 Prostate Cancer Version: 1.2023 Rectal Cancer Version: 2.2023 Small Bowel Adenocarcinoma Version: 1.2023 Small Cell Lung Cancer Version: 3.2023 Soft Tissue Sarcoma Version: 2.2023 Squamous Cell Skin Cancer Version: 1.2023 Systemic Light Chain Amyloidosis Version: 2.2023 Systemic Mastocytosis Version: 2.2022 T-Cell Lymphomas Version: 1.2023 Testicular Cancer Version: 1.2023 Thymomas and Thymic Carcinomas Version: 1.2023 Thyroid Carcinoma Version: 1.2023 Uterine Neoplasms Version: 2.2023 Vulvar Cancer Version: 1.2023 Waldenström Macroglobulinemia / Lymphoplasmacytic Lymphoma Version: 1.2023 Wilms Tumor (Nephroblastoma) Version: 1.2023 © National Comprehensive Cancer Network, Inc. 2023. All rights reserved. Accessed May 8, 2023. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.nccn.org). The NCCN Guidelines® and other content provided by NCCN are works in progress that may be refined as often as new significant data becomes available. They are statements of consensus of its authors regarding their views of currently accepted approaches to treatment. Any clinician seeking to apply or consult any NCCN Guidelines® or other NCCN Content is expected to use independent medical judgment in the context of individual clinical circumstances to determine any patient's care or treatment. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way. Therapeutic options are not applicable in all disease settings.

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VARIANT DETAILS - BIOLOGICALLY RELEVANT



NF1

c.7869+1G>A NM_001042492 Splice region variant - LOF

VAF: 0.4%

NF1 is a tumor suppressor that plays a role in cellular growth and differentiation through the regulation of the Ras protein. Loss of function mutations and copy number loss of NF1 are associated with cancer progression.

中文翻譯僅供參考，非正式報告

基因名稱：NF1

變異種類：剪接區域突變、功能喪失型突變

變異位點：c.7869+1G>A NM_001042492

變異等位基因比例：0.4%

基因功能解釋：NF1基因是我們DNA的一部分，它含有製造一種特殊蛋白質的指令。這種蛋白質有助於控制另一種蛋白質並參與細胞生長和分裂過程。當NF1基因正常運作時，會形成一種天然防禦機制防止癌症發生，透過調節來防止細胞不受控制地生長。然而，當NF1基因存在問題，例如基因突變或缺失時，會使細胞無法受到控制地生長和分裂，從而導致癌症的發生。(英文原譯: NF1是一種腫瘤抑制因子，透過調節Ras蛋白質在細胞生長和分化方面發揮作用。NF1的功能缺失突變和拷貝數缺失與癌症進展有關)。

TEMPUS 基因定序專有名詞解釋

Tempus 報告

當Tempus收到腫瘤標本，會對樣本進行檢測分析，大約2-3週後醫生會收到為每個患者準備的完整全面報告，重點突出關鍵發現，包括潛在可行的治療方法、免疫療法標誌物和匹配的臨床試驗。以下是報告中可能包含的專有名詞的解釋說明。



PDF檔案使用容易解說與理解的排版，適合醫師和病人共同閱讀和討論。另外提供TEMPUS專業版HUB，僅開放給醫師註冊使用。<https://accounts.securetempus.com/>

- 可排序和設定條件篩選全部報告列表
- 可連結到ClinicalTrials.gov臨床試驗數據庫
- 跟免疫治療相關的科學資訊(研究用途、僅限xT檢測才有)
- 可直接與TEMPUS Medical Affairs進行Case Review

基因變異

GENOMIC VARIANTS	
Somatic - Potentially Actionable	
CDKN2A p.A165 Frameshift - LOF	Variant Allele Fraction 40.7%
KRAS p.G12V Point mutation - GOF	22.0%
BRC2 Copj Number Loss - LOF	
Germline	
BRC2 p.S611* chr13:32907447	Clinical Significance Pathogenic Hereditary breast and ovarian cancer
TPMT p.A80P chr6:18143724	Pharmacogenetic Variant
No reportable single nucleotide variants, indels, or copy number changes found in: TP53 SMAD4	

當我們發現癌細胞裡的DNA變異，就稱它為「體細胞基因變異」，是癌症發生的原因。有些「體細胞基因變異」可以有標靶藥物，但不是所有基因變異都有標靶藥物，原因可能是：1.突變位點並非用藥對應位點、2.或是藥物還在臨床試驗階段、3.或是藥物還在研發階段。

體細胞

如果這個基因變異有標靶藥物或療法，可以減緩或阻止癌細胞的生長和擴散，或縮小腫瘤，或是可搭配其他藥物提高治療效果；或是已知會讓某種標靶藥物產生抗藥性，那麼這個基因變異就被定義是「潛在可行動的變異」

潛在可行動的變異

體細胞

如果這個基因變異，以目前的科學能力尚未開發出可治療的藥物，那麼這個基因變異就定義是「生物學相關的變異」

生物學相關的變異

生殖細胞變異 僅限 xT 檢測才有

這些是跟遺傳有關的突變，有的跟罹癌風險有關，有的跟美國醫學遺傳學和基因學院 (ACMG) 所列表之「醫學上可採取行動的」的基因有關



MUTATIONS 突變

突變是基因DNA序列的變化，可以在體細胞（後天性得到的）或生殖細胞（先天遺傳的）。此外，對於每個體細胞突變，Tempus提供變異等位基因比例（VAF），即定序檢體中基因組變異的讀取比例，以百分比表示。



COPY NUMBER VARIATIONS (CNV) 拷貝數變異

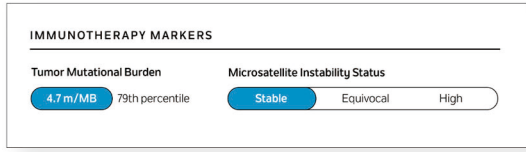
基因拷貝數變異報告有2種：包括擴增（拷貝數增加）或缺失（拷貝數減少）報告。Tempus透過DNA定序檢測CNV。若是失去雜合性（LOH）失去兩個編碼等位基因中的一個，也會在報告中註明。



CHROMOSOMAL REARRANGEMENTS (TRANSLOCATIONS) 染色體重組(易位)

染色體結構重組是指兩個分開的基因片段重組在一起形成融合基因。融合基因可能會產生異常和/或過度活躍的蛋白質，進而導致腫瘤生成、進展或抗藥性。Tempus 採用兩種序列檢測方法檢測重組（易位）：對部分基因進行DNA序列檢測，以及對全轉錄組RNA序列進行融合分析(RNA融合分析僅限xT檢測才有)。

免疫治療生物標記



檢測免疫治療的關鍵生物標記，包括微衛星不穩定狀態 (MSI)、腫瘤突變負荷 (TMB)，以及 PD-L1 和 MMR 錯配修復 (PD-L1 和 MMR 是選購項目，若無選購則無此報告)

TUMOR MUTATIONAL BURDEN (TMB) 腫瘤突變負荷量

腫瘤突變負荷 (TMB) 是一個衡量腫瘤中攜帶的突變數量的數值，即每百萬個鹼基對中改變蛋白質的體細胞突變的數量。研究顯示，TMB 高的腫瘤對免疫療法的反應可能性增加。TEMPUS 的 TMB 報告是通過將非同義突變數量除以 TEMPUS Panel 的每百萬個鹼基數計算得出的。將 TMB 大於等於 10 定義為“高”。除了 TMB 原始分數 (mut/Mb)，TMB 百分位數是報告中提供的另一個可用數據，代表檢測樣本的 TMB 分數在整個 Tempus 癌數據庫中的排名。

MICROSATELLITE INSTABILITY (MSI) 微衛星不穩定性

微衛星不穩定性 (MSI) 狀態是指因 DNA 錯配修復受損而導致的基因組不穩定性，是一種免疫治療生物標記物參考。Tempus 採用 DNA 定序分析 MSI 狀態 (也可選購 IHC-MMR)。微衛星不穩定性-高 (MSI-H) 代表與對免疫療法反應的可能性增加有關。

治療意義

FDA-APPROVED THERAPIES, CURRENT DIAGNOSIS	
PARP Inhibitor	Olaparib
BRCA2 Loss-of-function Consensus, ovarian cancer: FDA	

根據這次送檢的腫瘤基因分析報告，提出有醫學證據的治療方法

- ✓ 美國 FDA 核可療法，適用目前診斷
- ✓ 美國 FDA 核可療法，其他適應症
- ▲ 研究性療法是指正在進行臨床試驗研究和評估，尚未被監管機構批准使用的醫療方法。臨床試驗階段確定其安全性、有效性、劑量和潛在的副作用。研究性療法通常被使用在已用盡所有可用治療方案，或用於那些患有罕見或難以治療的疾病的人。參加研究性療法臨床試驗的病人由醫療服務提供者密切監測，以評估該療法的潛在益處和風險。

臨床試驗

CLINICAL TRIALS	
Genetic analysis-guided dosing of FOLFIRI in treating patients with locally advanced or metastatic pancreatic cancer	Phase I/II NCT02688855 University of Chicago, Icahn
Genetic analysis-guided dosing of FOLFIRI in treating patients with advanced gastrointestinal cancer	Phase I/II NCT02688855 University of Chicago, Icahn
Study of PD-1 Blockade by Pembrolizumab With Stereotactic Body Radiotherapy in Advanced Solid Tumors	Phase I NCT02688855 University of Chicago, Icahn

根據此次送檢的腫瘤基因特徵以及癌症類型和臨床病史，TEMPUS 為您配對了一份最新的臨床試驗清單

意義不明的變異

VARIANTS OF UNKNOWN SIGNIFICANCE		
Somatic variant	Mutation effect	Variant allele fraction
ARHGAP5	c.1441G>T p.E481*	18.6% —
ARHGAP5	c.1465G>A p.E489K	22.1% —
CCT6A	c.725+1G>A Splice-Donor	21% —

意義不明的體細胞變異 (VUS) 是指這個 DNA 變異與癌症生物學有關的證據還不夠充分或證據還不夠明確。為了避免與“生物相關”和“臨床可操作”混淆，TEMPUS 不會報告意義不明的變異 (VUSs) 的基因變異。

低覆蓋區域

LOW COVERAGE REGIONS				
CDKN1C	GFRA2	NOTCH1	PDPK1	RECQL4
FLT4	MTAP	NOTCH2	PIK3R2	ZNRF3

低覆蓋率區域是指基因被定序的深度或質量低於實驗室的閾值規定。由於技術限制或其他因素，某個區域沒能以符合實驗室規定的深度或質量被定序。低覆蓋率區域會使得基因變異鑑定變得困難，所以若確實有變異也可能無法被檢測出。

變異詳細說明

SOMATIC VARIANT DETAILS - POTENTIALLY ACTIONABLE

C: CDKN2A c.225delC p.A376s Frameshift - Loss-of-Function Variant Allele Fraction: 40.7%

CDKN2A encodes two proteins, p16^{INK4A} and p14^{ARF} which function in regulating cell growth. The p16^{INK4A} protein regulates the cell cycle through the inhibition of CDK4 and CDK6, preventing them from stimulating cell proliferation. The p14^{ARF} protein binds to MDM2 to keep p53 inhibited and stimulate the p53-dependent cell cycle arrest and apoptosis. Deleterious single nucleotide variants, copy number loss and underexpression of CDKN2A are associated with cancer progression.

Tempus採用次世代定序檢測體細胞可操作和生物相關的基因組變異和/或偶然發現的生殖細胞突變。Tempus的報告總結了檢測到的基因變異，包括變異位置、影響和基因功能。

- 錯義突變：** 錯義突變是一種可能發生在我們DNA中的變化。我們的基因是由DNA組成的，DNA包含構建我們身體正常運作所需的所有蛋白質的指令。當DNA編碼指令的一個代碼發生變化時，就會發生錯義突變，進而導致產生不同的氨基酸，進而影響蛋白質的功能，這種改變可能導致疾病或根本沒有影響。錯義變體是否有害取決於許多因素，如變化的位置和它影響的蛋白質的重要性。
- 終止型突變：** 終止型突變是一種發生在基因中的突變，這種突變導致了一個終止密碼子的產生，它會讓蛋白質合成提早結束，並生成為一個斷掉不完整的蛋白質，這個蛋白質通常是無功能或功能障礙的。這種類型的突變有時也被稱為無義突變或過早終止密碼子突變。
- 剪接區域突變：** 剪接位點是基因的特定區域，在這裡DNA序列被切割並粘貼在一起，以形成最終的mRNA，用於構建蛋白質。這個過程被稱為剪接。然而，如果剪接位點出現突變，剪接過程可能出錯，基因的外顯子（編碼區）可能無法正確地剪接在一起。其結果是往往是一個外顯子被排除在之外，這可能導致產生一種異常的蛋白質或根本沒有蛋白質。
- 功能獲得型突變：** 用於描述導致蛋白質具有增強或新功能的基因變化。當一個突變改變了一個基因的DNA序列，導致其編碼的蛋白質變得更有活性、或是出現新的功能。
- 功能喪失型突變：** 導致蛋白質失去正常功能的DNA序列變化。
- 移碼突變：** 移碼突變是一種基因突變，通常是由於基因序列中的插入或刪除等錯誤而導致的，會改變蛋白質翻譯的讀框，從而使其產生不同的氨基酸序列，並可能導致蛋白質合成的提前終止。通常會導致蛋白質喪失正常功能。
- 內部缺失突變：** 內部缺失突變是基因突變的一種，其中一小段DNA缺失(通常是3個或3的倍數)，這會導致基因的錯誤拼接，但是整個基因依然能夠翻譯成一個功能性的蛋白質。換言之，可以想像成是拼圖少了一塊，但是整張圖還是能夠看出來。這種突變通常會影響基因的功能，有時也會和癌症的發生有關。