GENETIC DISORDER, HPO, OMIM and ClinVar

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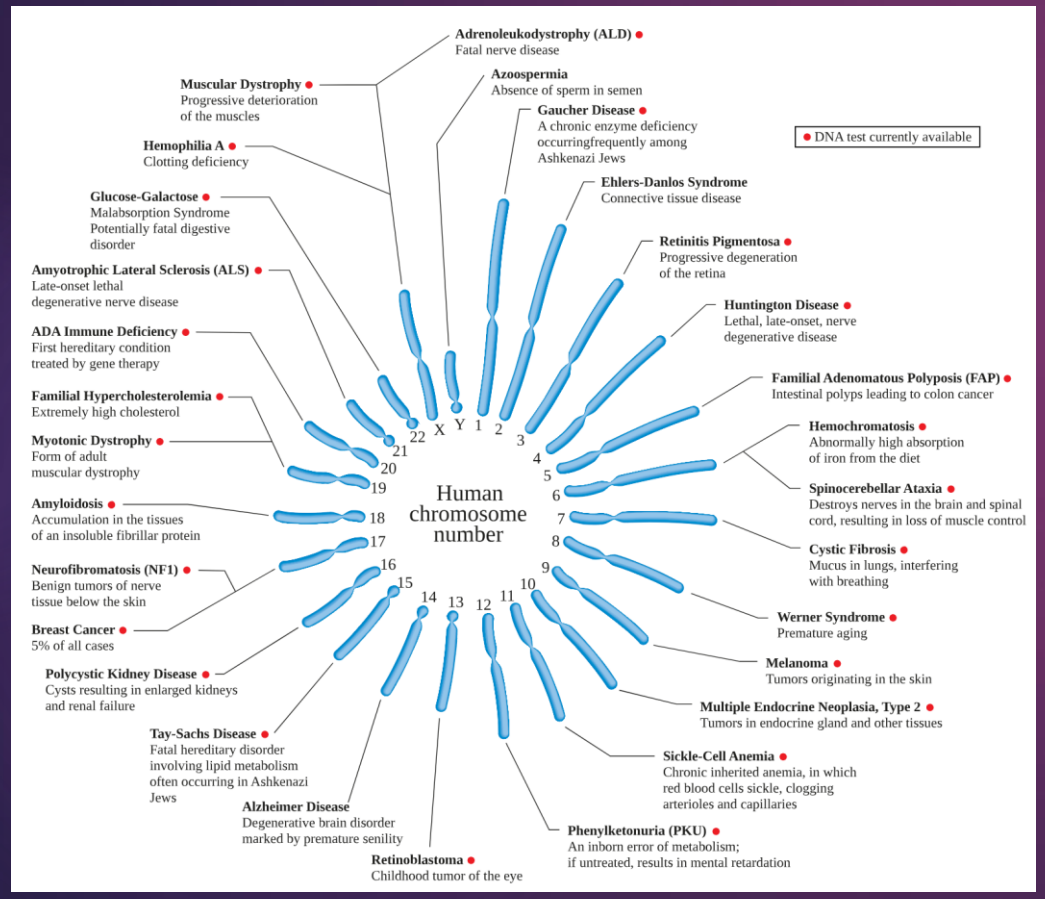
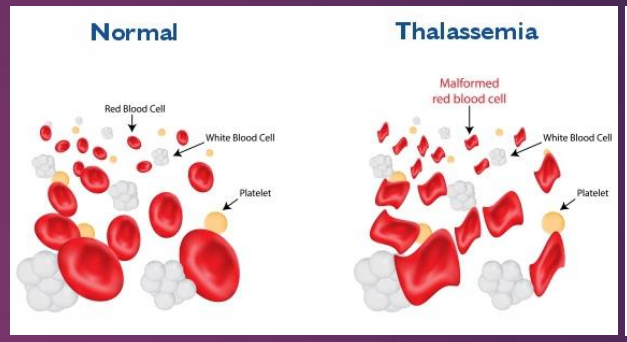
Content of Lecture 3

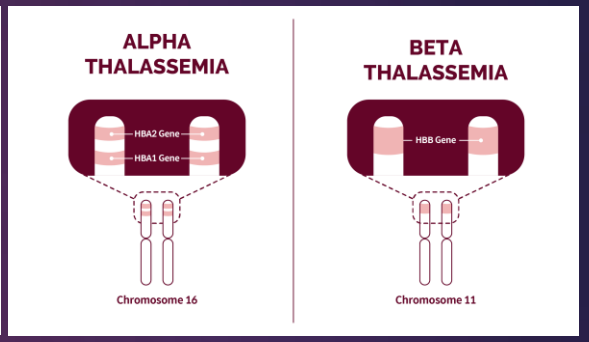
1. Genetic disorder
2. Introduction to HPO (The Human Phenotype Ontology)
3. Introduction to OMIM
4. Introduction to ClinVar

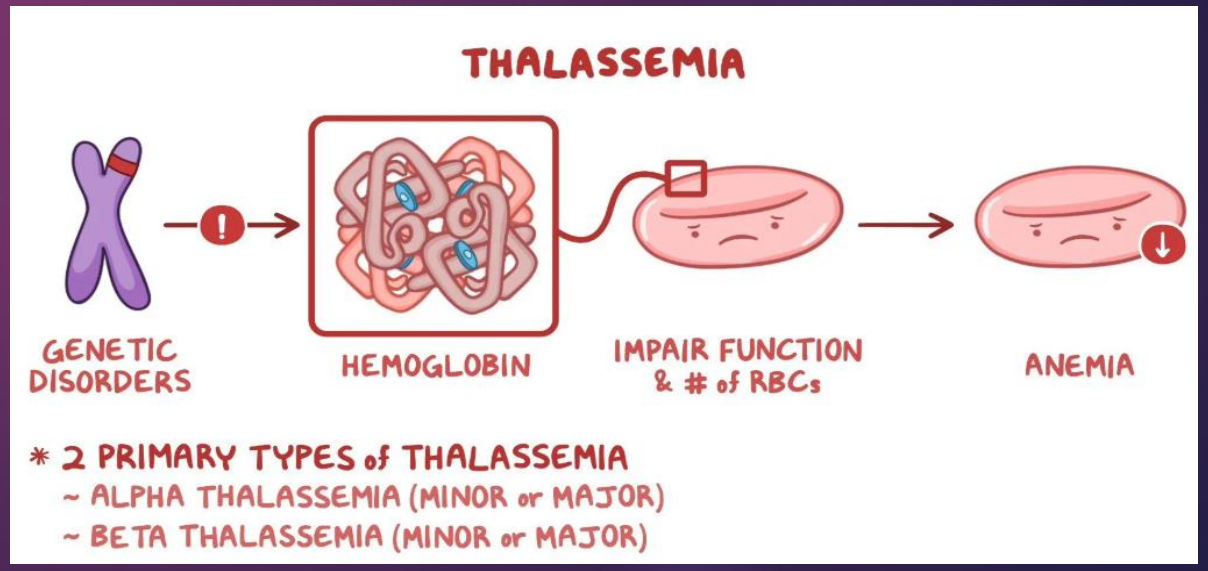
Genetic disorder

- A genetic disorder is a health problem caused by one or more abnormalities in the genome. It can be caused by a mutation in a single gene or multiple genes or by a chromosome abnormality.



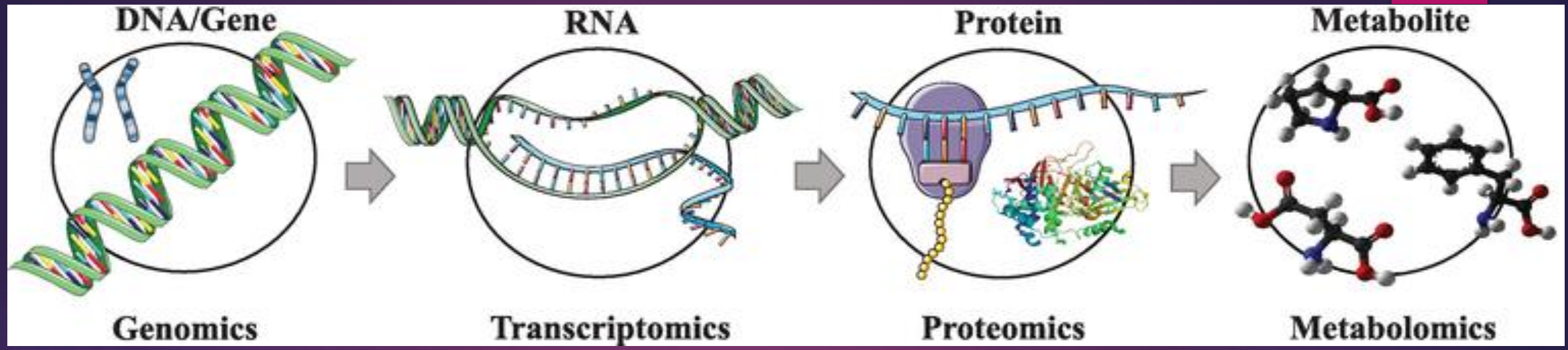






https://en.wikipedia.org/wiki/Genetic_disorder

Quan hệ nhân quả: Biến thể gen và bệnh di truyền



PAH gene

Ref …ATCGAT…

P1 …AACGAT…

NM_000277.3(PAH):c.971T>A

PAH mRNA

Ref …AUCGAU…

P1 …AACGAU…

NM_000277.3(PAH):c.971T>A

PAH protein

Ref …Ile-Asp…

P1 …Asn-Asp…

NM_000277.3(PAH):p.Ile324Asn

PAH

Ref Phe → Tyr

PAH

P1 Phe ↛ Tyr

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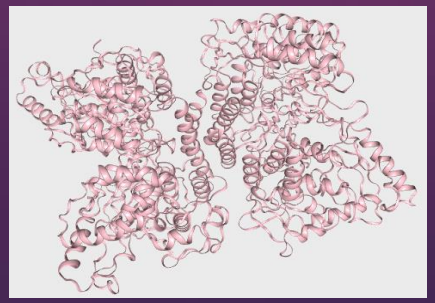

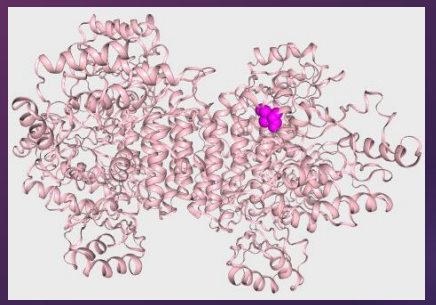

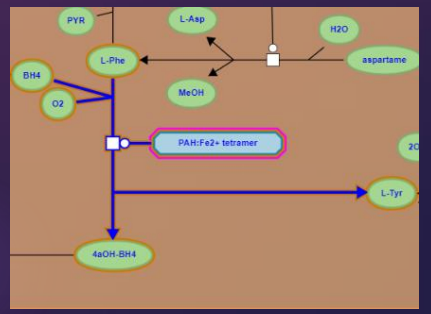

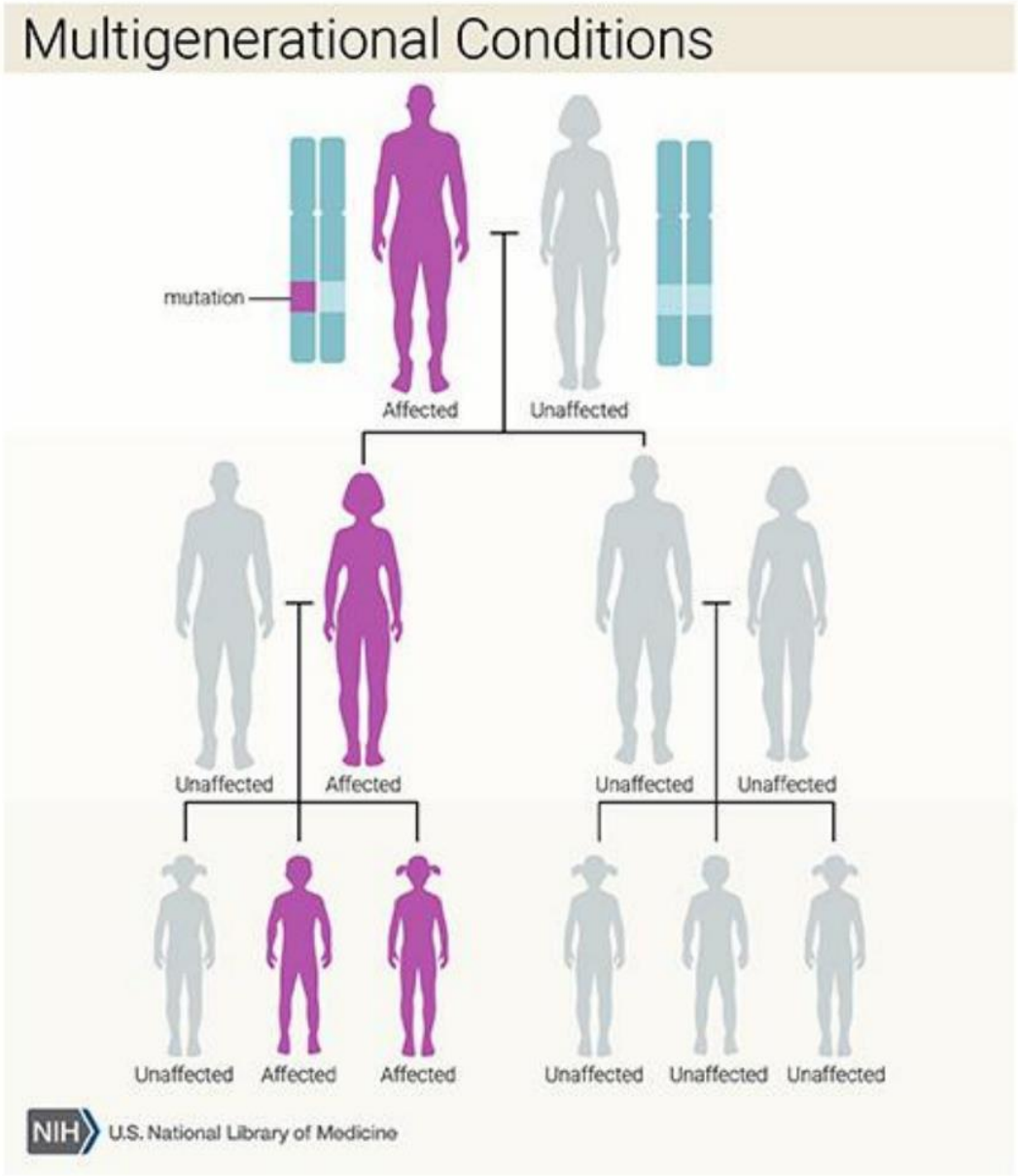

FIGURE 1: Three generations of a family with a genetic disorder.

https://medlineplus.gov/genetics/

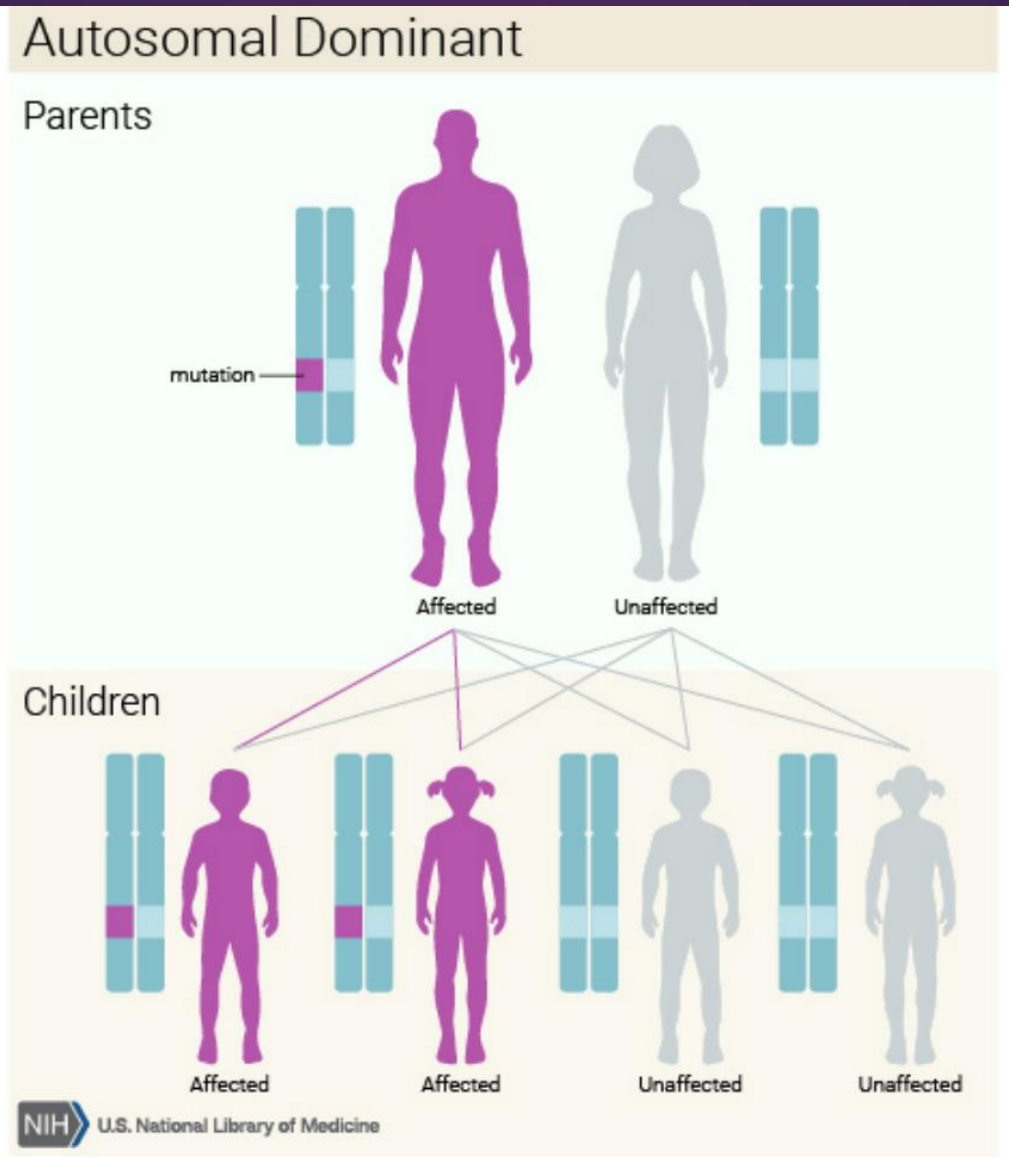

FIGURE 2: A parent with an autosomal dominant condition passes the altered gene to two affected children. Two other children do not receive the altered gene, and are unaffected.

https://medlineplus.gov/genetics/

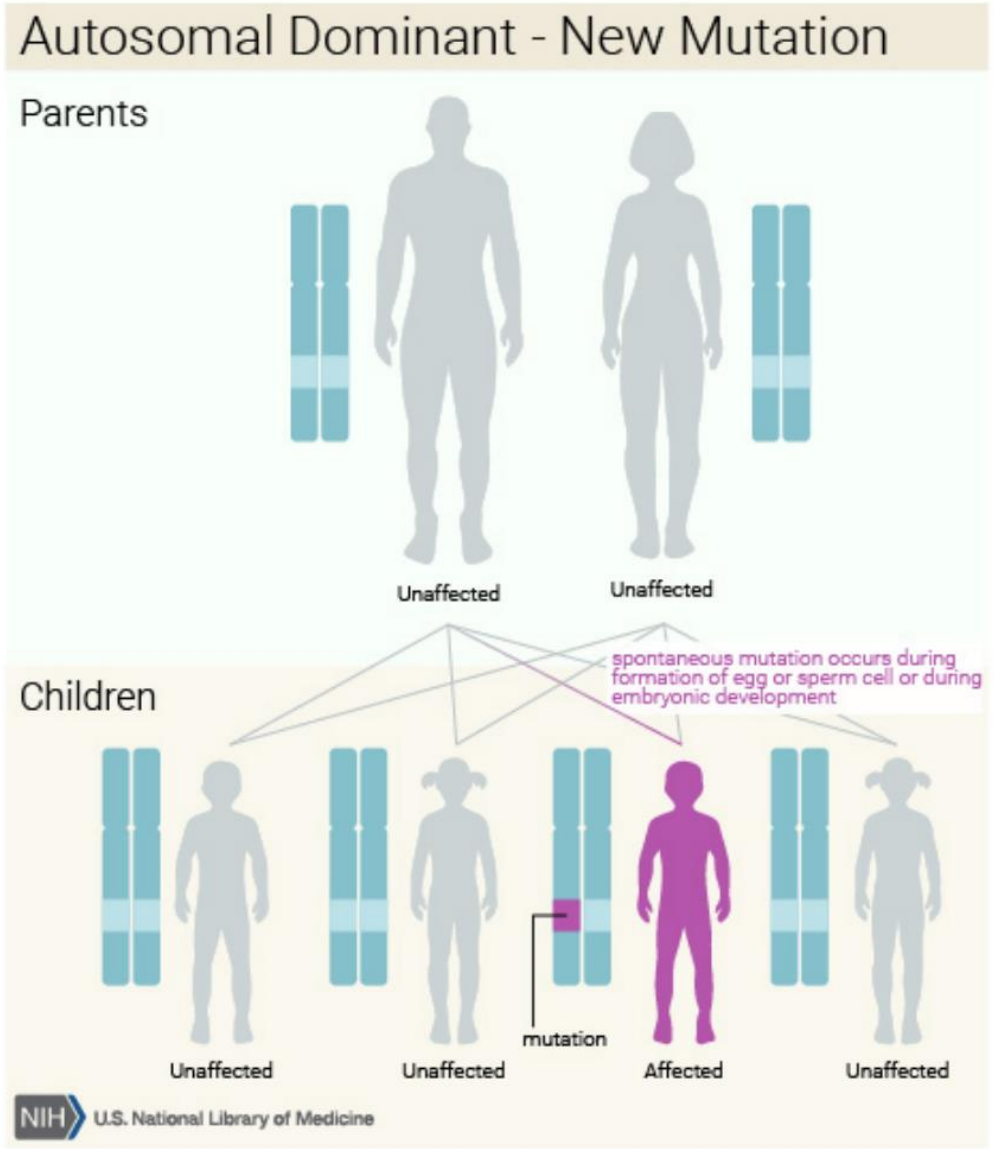

FIGURE 3: Neither parent has the mutated gene. A spontaneous mutation occurs during the formation of an egg or sperm cell during embryonic development, leading to an affected child.

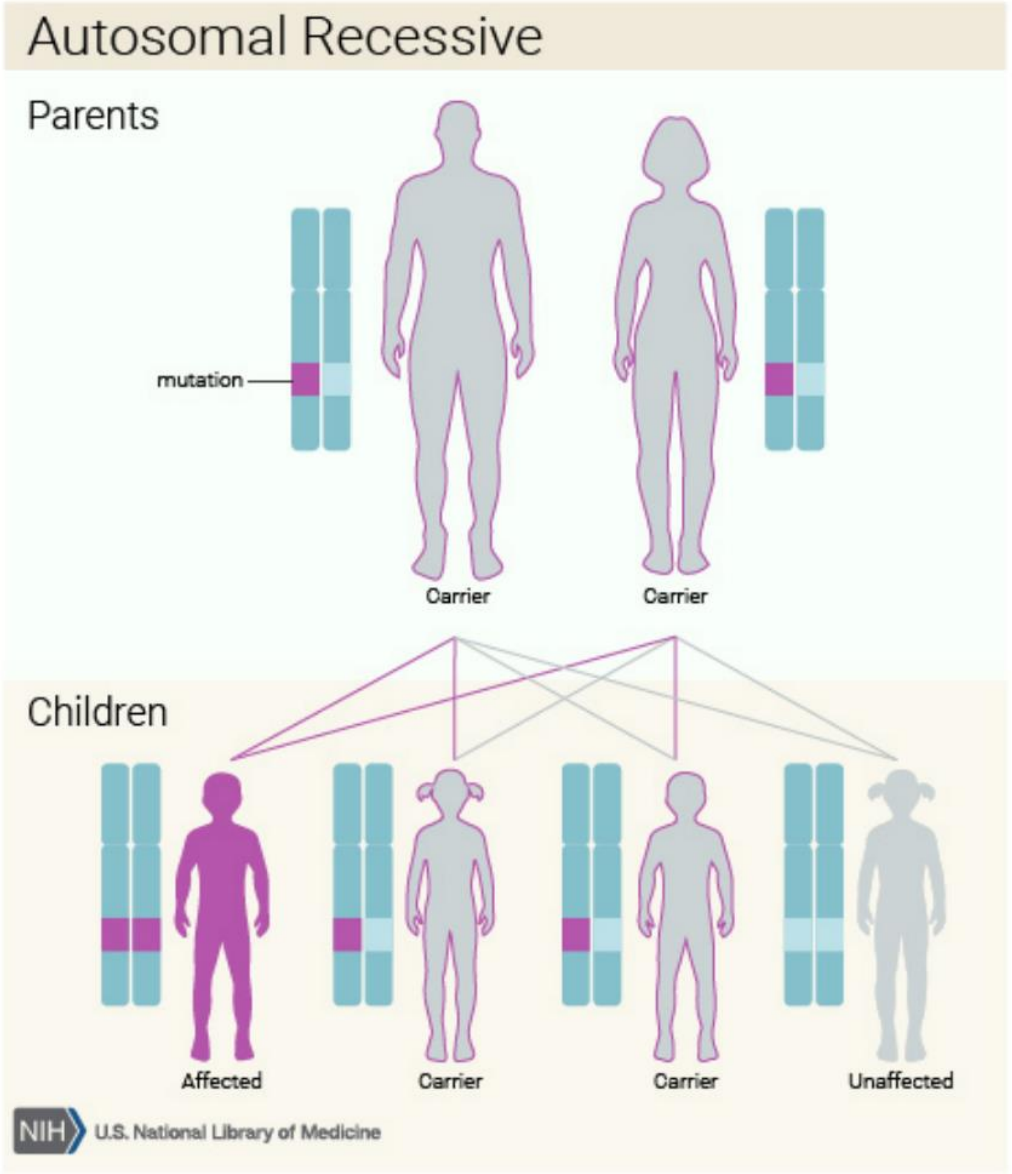

FIGURE 4: Both parents carry one copy of a mutated gene. In the next generation, one child is affected with the condition, two children are carriers, and one is unaffected and not a carrier.

https://medlineplus.gov/genetics/

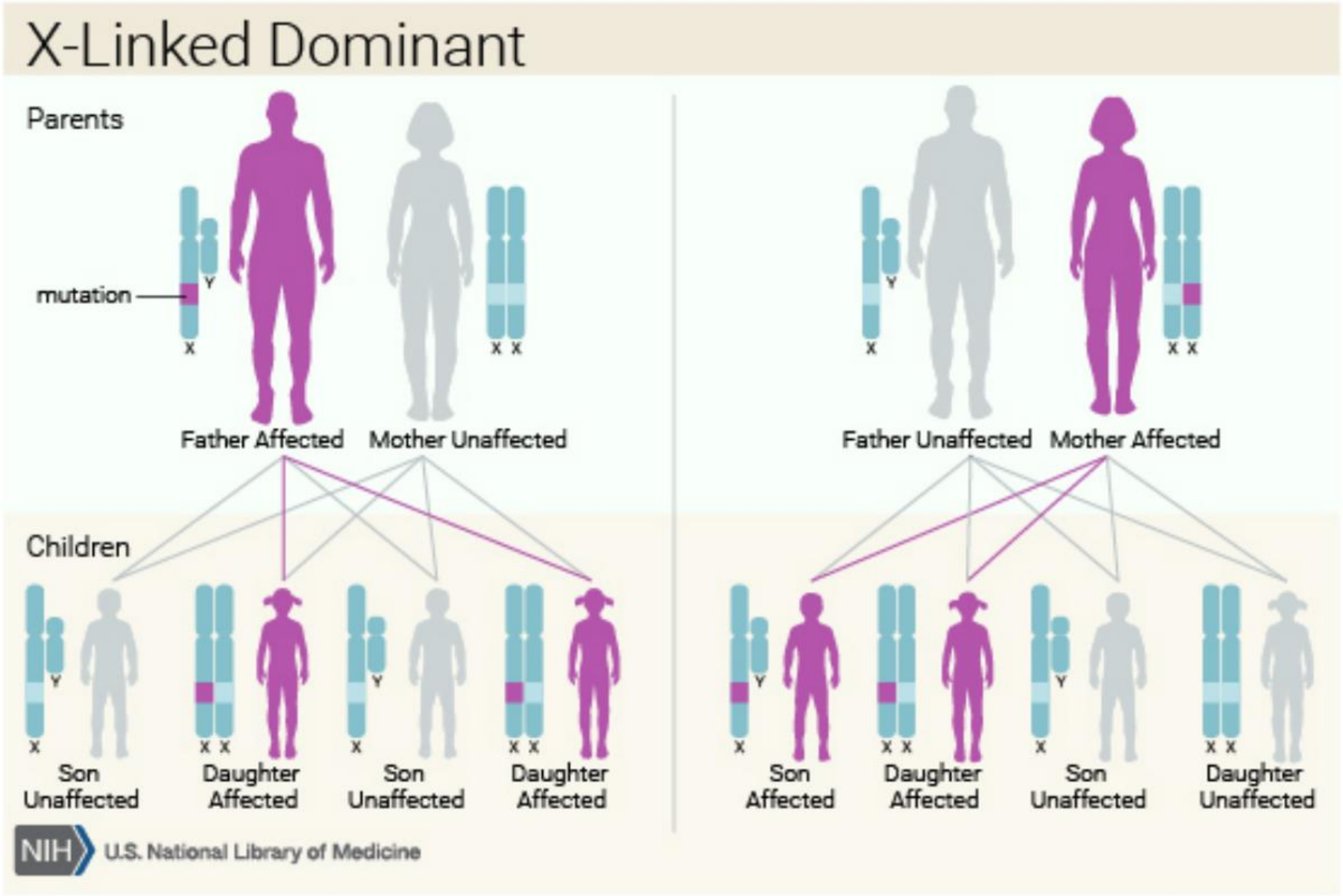

FIGURE 5: Inheritance of an X-linked dominant disorder depends on which parent is affected.

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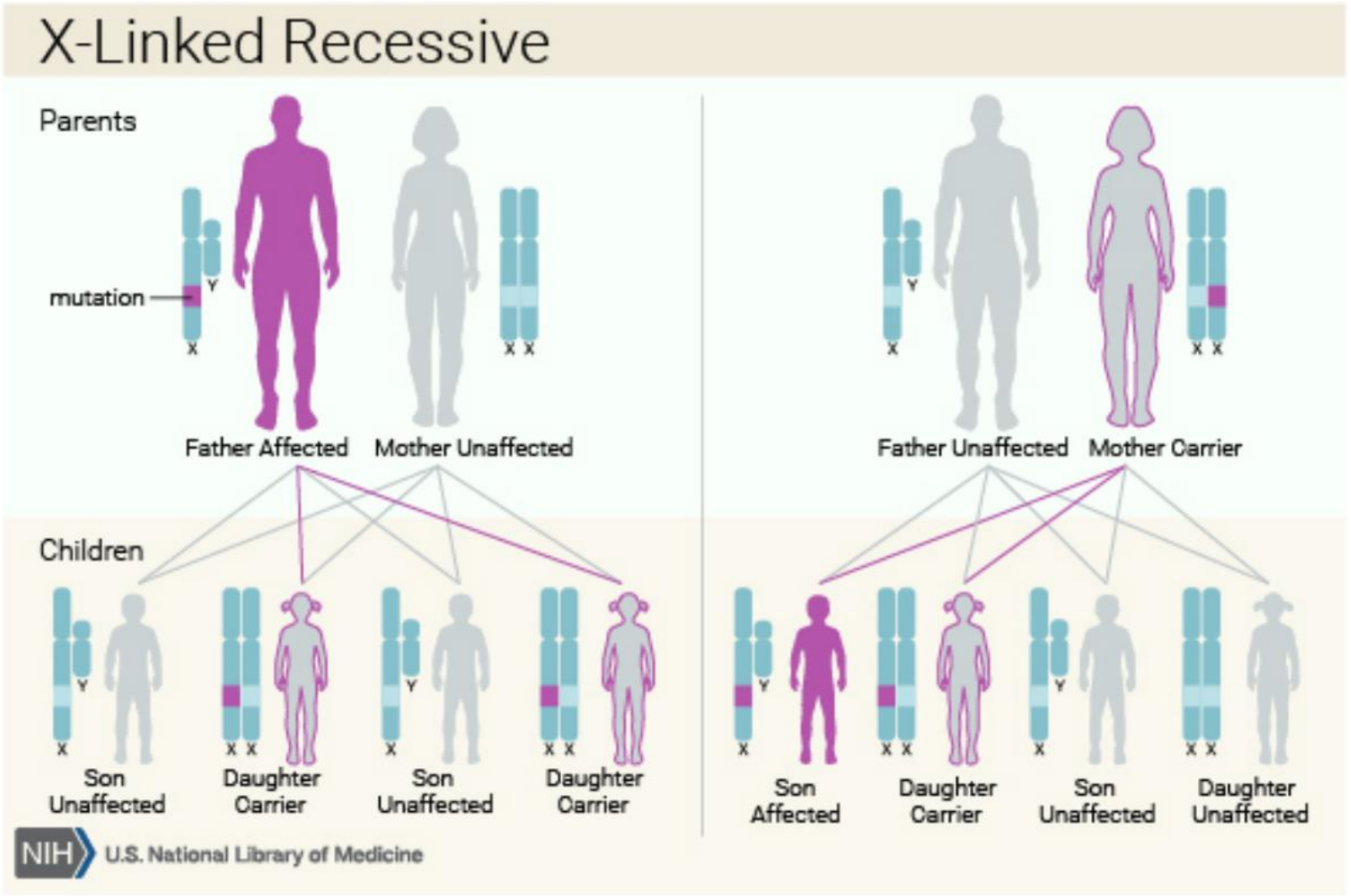


FIGURE 6: Two generations of a family with an X-linked recessive disorder. In this form of inheritance, the chance of being affected or being a carrier depends on whether the mother or the father has the mutated gene on the X chromosome.

eplus.gov/genetics/

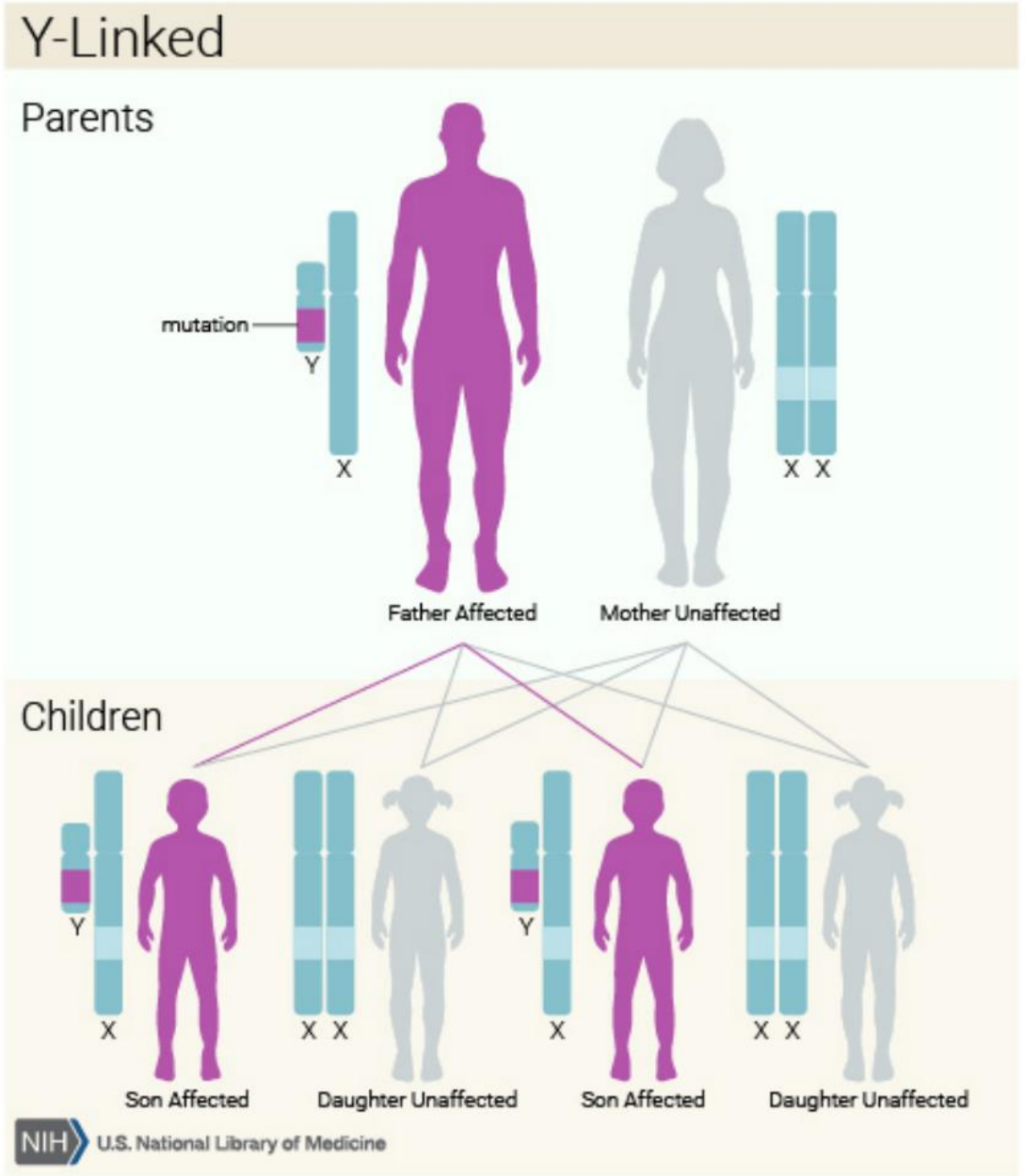


FIGURE 7: A father and sons are affected with a Y-linked disorder, which is

https://medlineplus.gov/genetics/

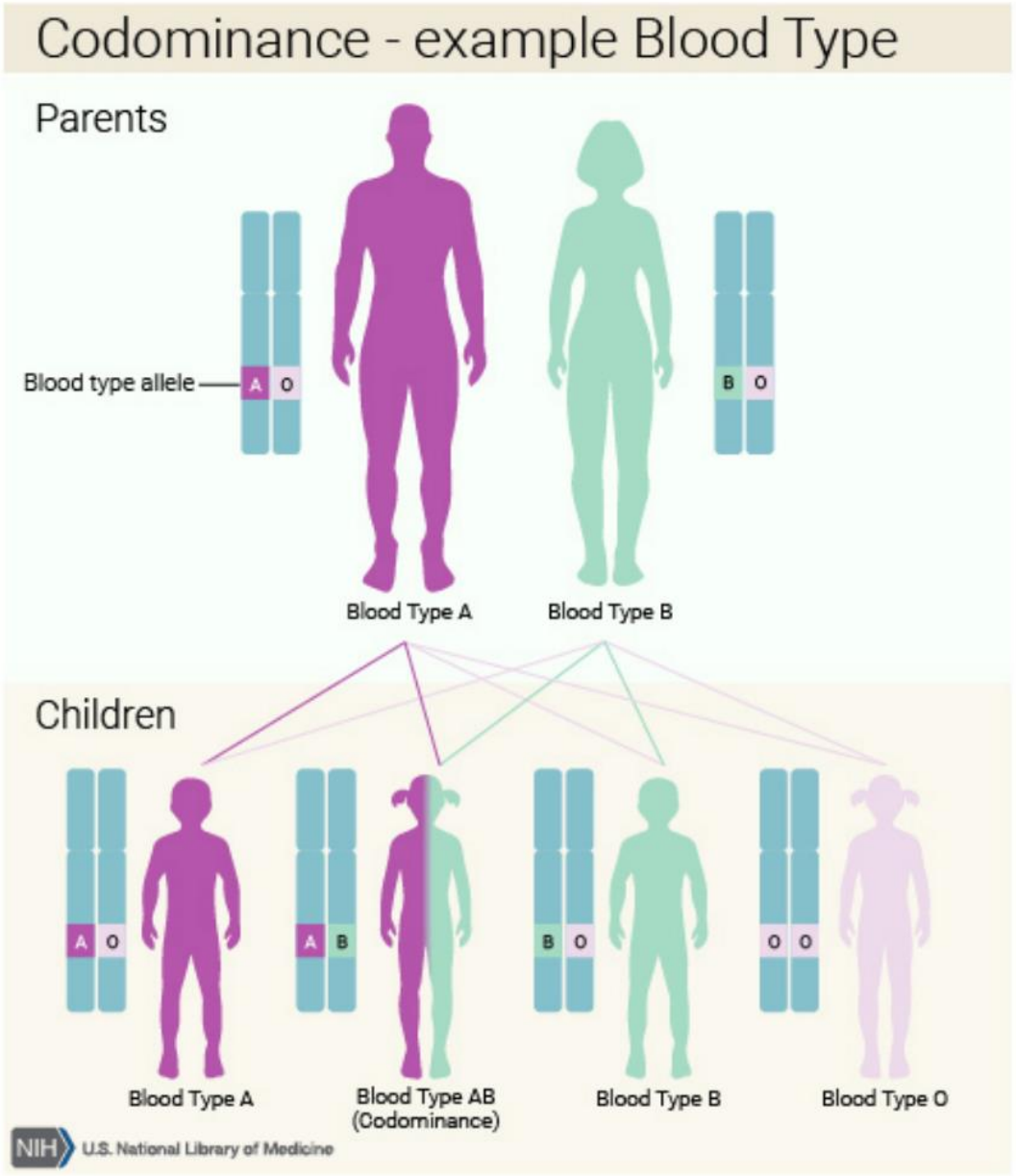


FIGURE 8: ABO blood type is an example of a trait with codominant inheritance.

https://medlineplus.gov/genetics/

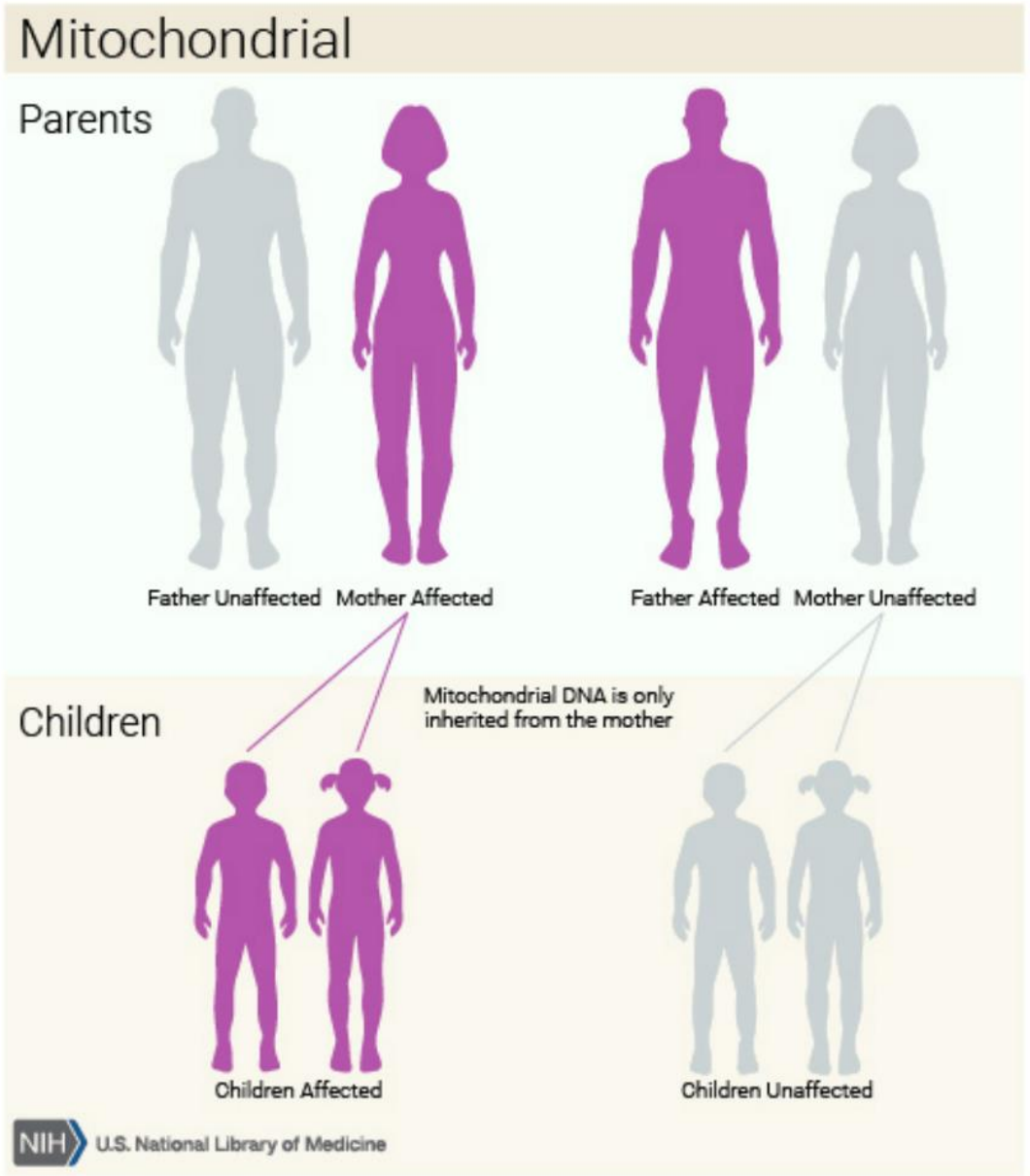
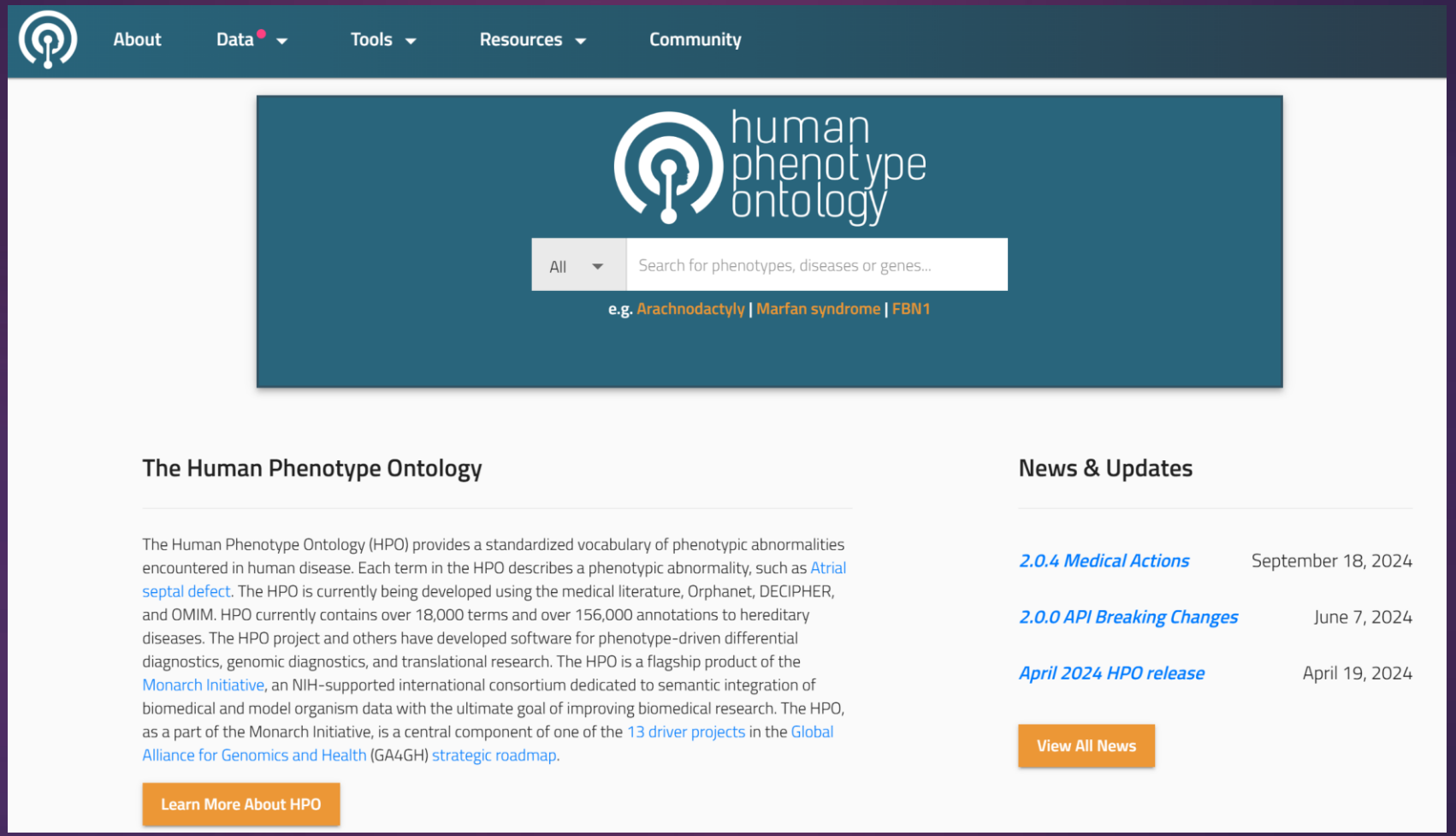


FIGURE 9: The inheritance of a mitochondrial disorder depends whether the mother or the father has the mutation in mitochondrial DNA.

https://medlineplus.gov/genetics/

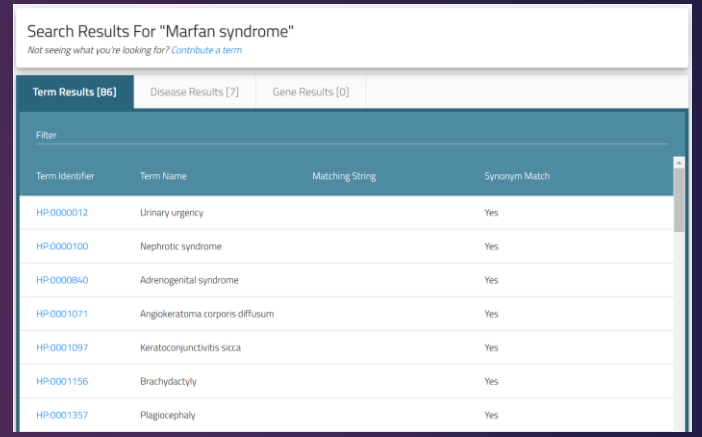
The Human Phenotype Ontology (HPO)

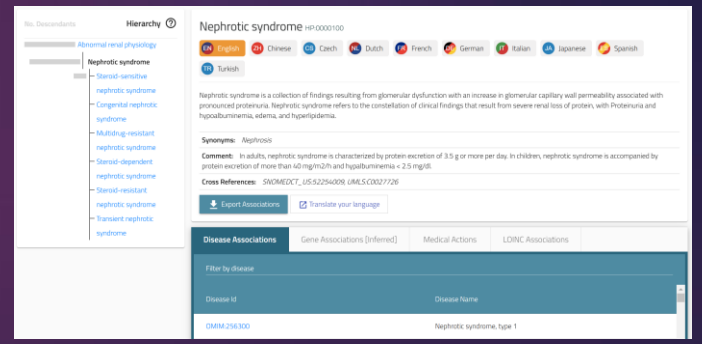


https://hpo.jax.org/

The Human Phenotype Ontology (HPO)

- The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease.
- Each term in the HPO describes a phenotypic abnormality, such as Atrial septal defect.
- The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM.
- HPO currently contains over 18,000 terms and over 156,000 annotations to hereditary diseases.
- The HPO project and others have developed software for phenotype-driven differential diagnostics, genomic diagnostics, and translational research.





https://hpo.jax.org/

The Human Phenotype Ontology (HPO)

Disease Associations | **Gene Associations [Inferred]** | Medical Actions | LOINC Associations

Filter by gene

Gene Id	Gene Symbol
NCBIGene:4868	NPHS1
NCBIGene:112858	TP53RK
NCBIGene:2013	EMP2
NCBIGene:286204	CRB2
NCBIGene:9688	NUP93
NCBIGene:9863	MAGI2

139 gene associations.

https://hpo.jax.org/browse/term/HP:0000100

The Human Phenotype Ontology (HPO)

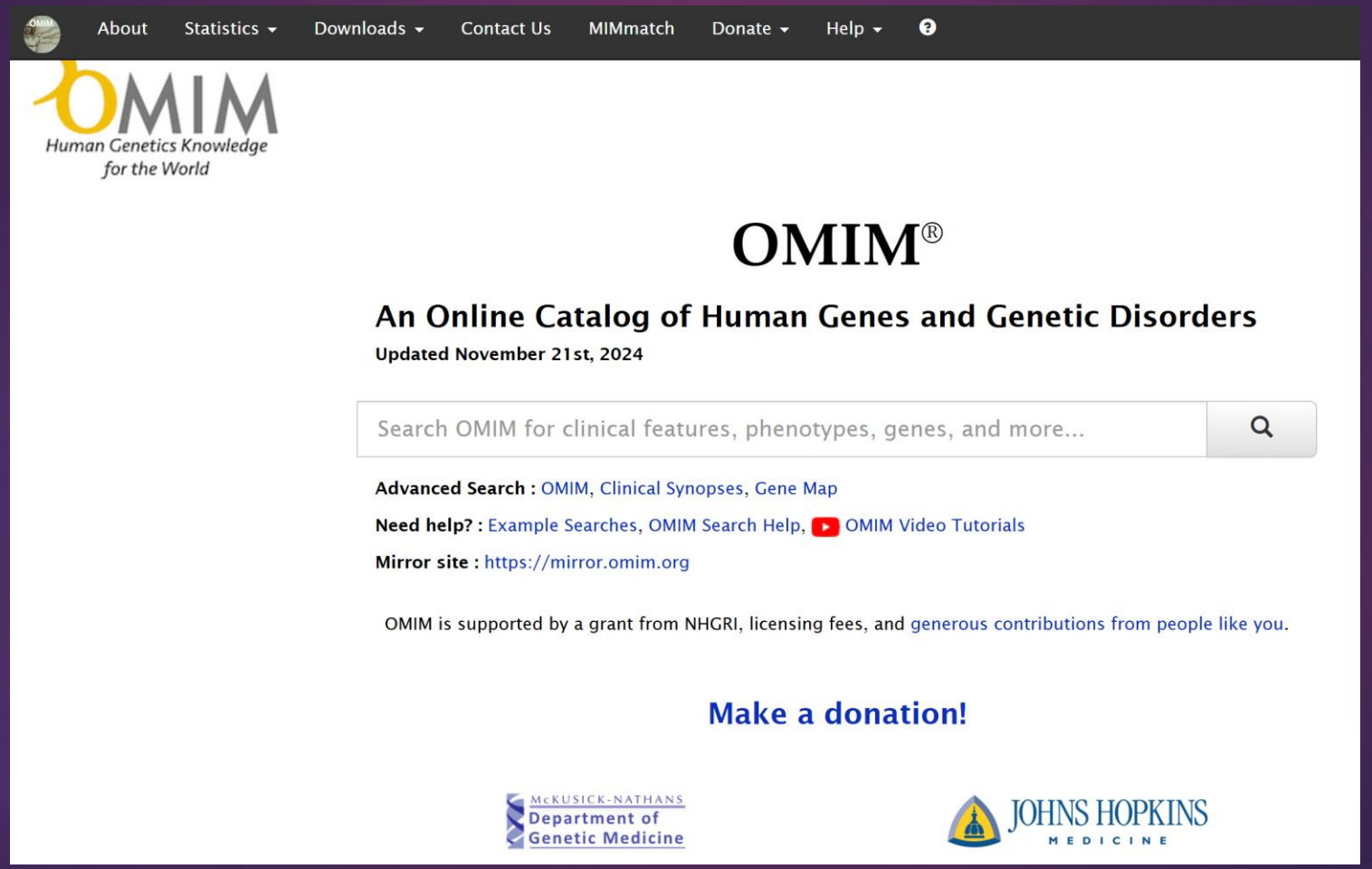
Disease Associations | Gene Associations [Inferred] | **Medical Actions** | LOINC Associations

MaXo Id	MaXo Name	Relation	Sources
MAXO:0000653	angiotensin receptor blocker therapy	TREATS	PubMed
MAXO:0000640	corticosteroid agent therapy	TREATS	PubMed
MAXO:0000297	immune suppressant agent therapy	TREATS	PubMed
MAXO:0000190	RAAS inhibitor therapy	TREATS	PubMed
MAXO:0000652	ACE inhibitor therapy	TREATS	PubMed

5 medical actions.

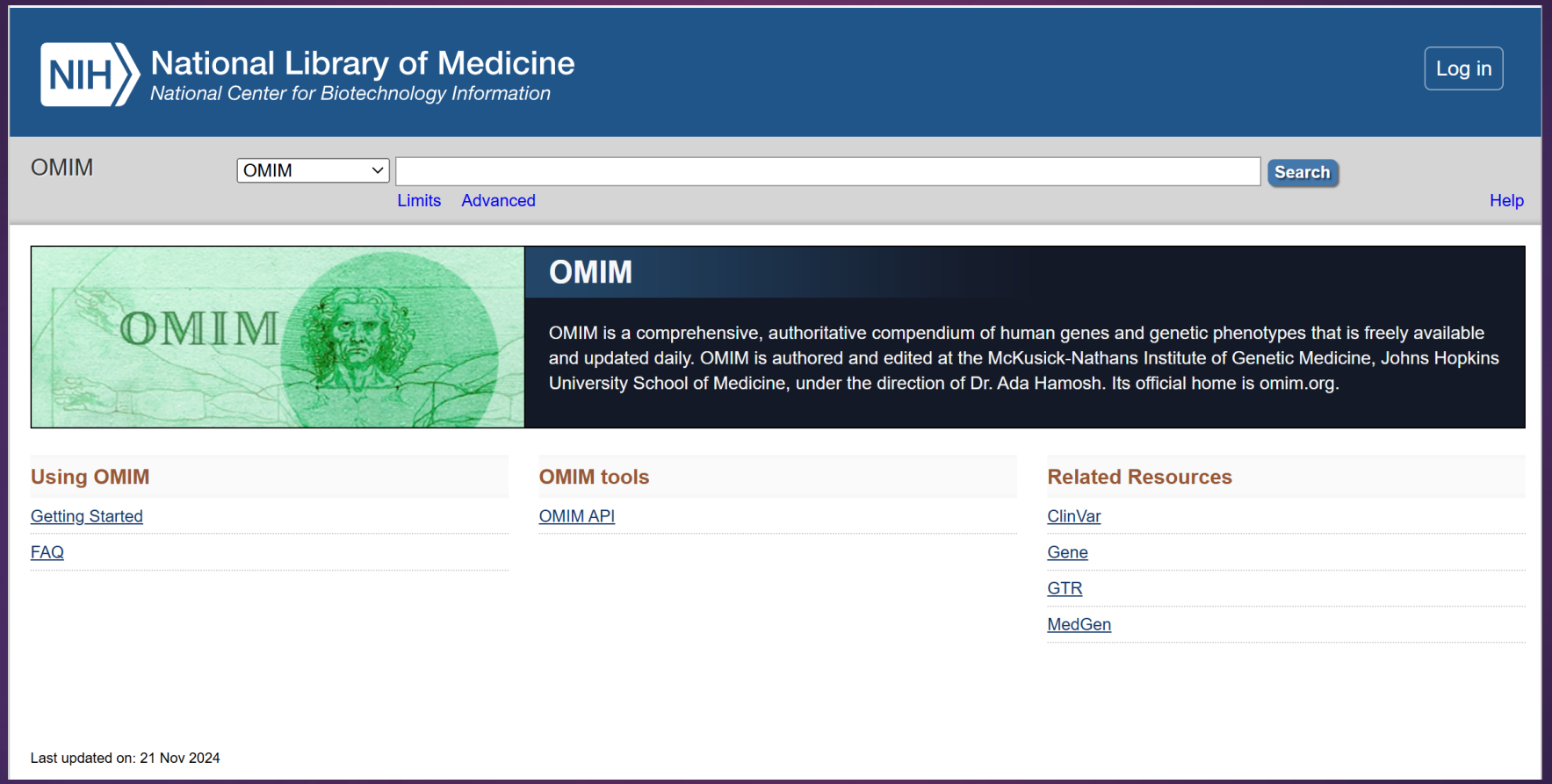
https://hpo.jax.org/browse/term/HP:0000100

OMIM: An Online Catalog of Human Genes and Genetic Disorders



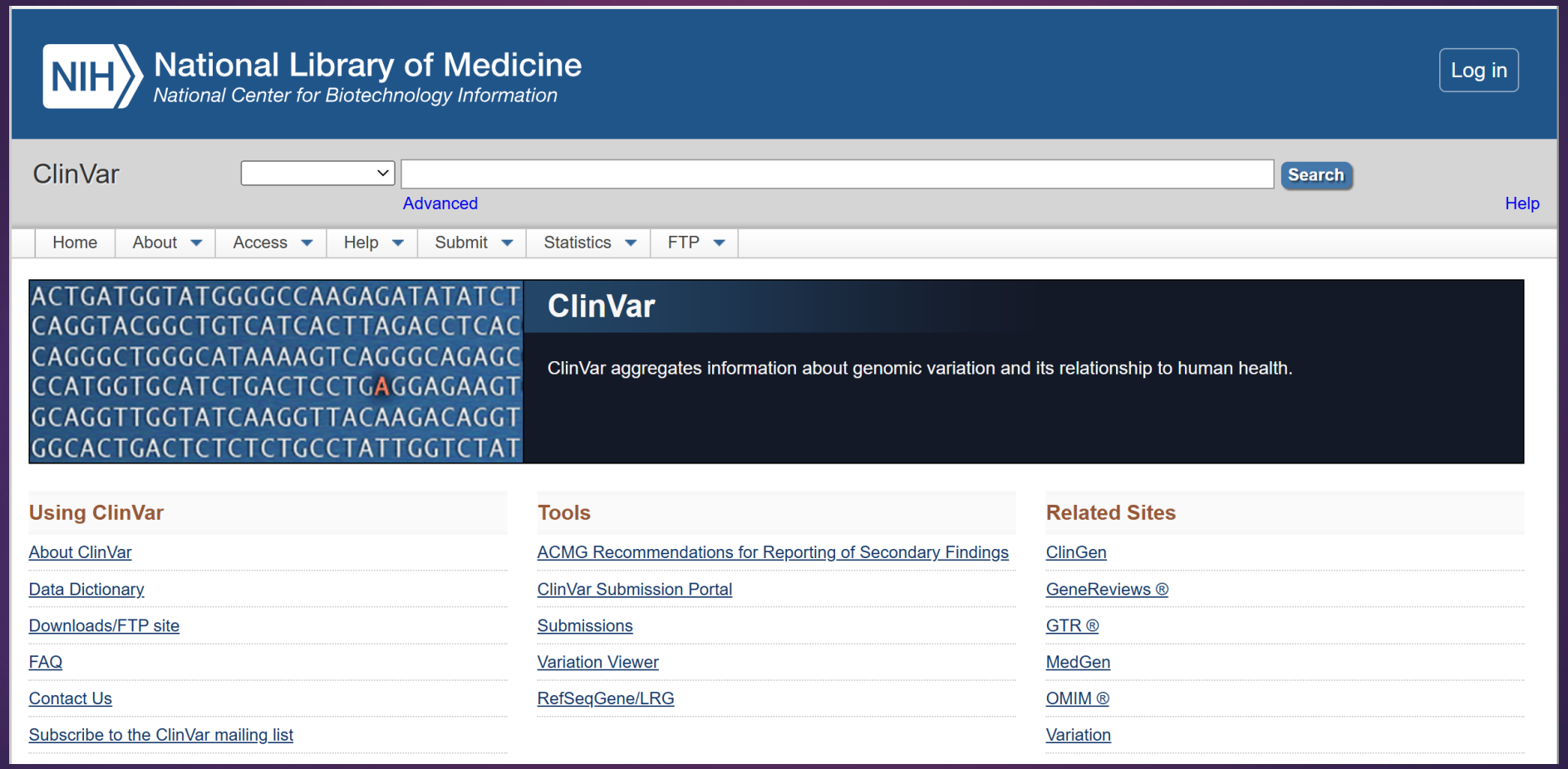
https://omim.org/

NCBI OMIM: An Online Catalog of Human Genes and Genetic Disorders



https://www.ncbi.nlm.nih.gov/omim

ClinVar: Reports of human variations classified for diseases and drug responses



https://www.ncbi.nlm.nih.gov/clinvar/

Xin chân thành cảm ơn!

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