



Genomic discovery

“Why is it this way?”

~~“Because we say so”~~

Our doctor is an expert knowledge curator.

Provide medical diagnosis
that is evidence-based.

How can I trust medicine from
an algorithm?

[Video clip: Computers can not
replace AT&T switchboard operators](#)

Artificial Intelligence—The Revolution Hasn’t Happened Yet

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<https://doi.org/10.1162/99608f92.f06c6e61>

Related PopSci “**Stop Calling Everything AI, Machine-Learning Pioneer Says**”

<https://spectrum.ieee.org/stop-calling-everything-ai-machinelearning-pioneer-says>

We can explain your illness.

Due to a variant in your DNA,
your immune systems is often overactive.

This causes inflammation and pain.

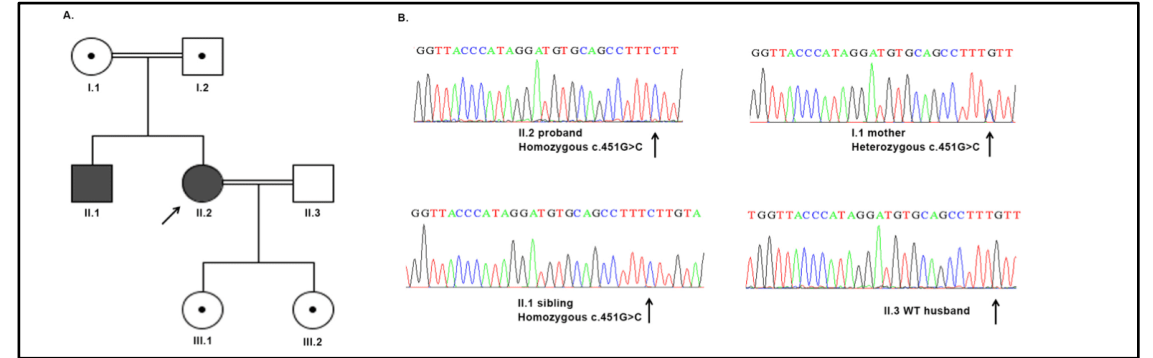
We know the mechanism because of
existing scientific evidence.

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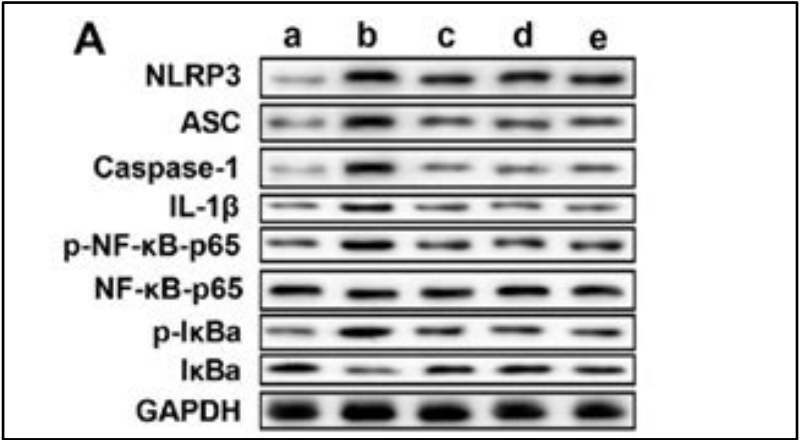
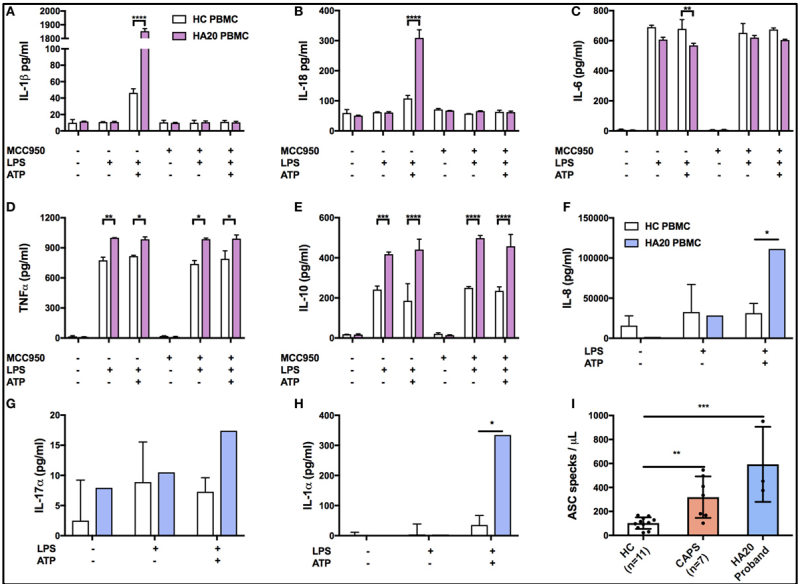


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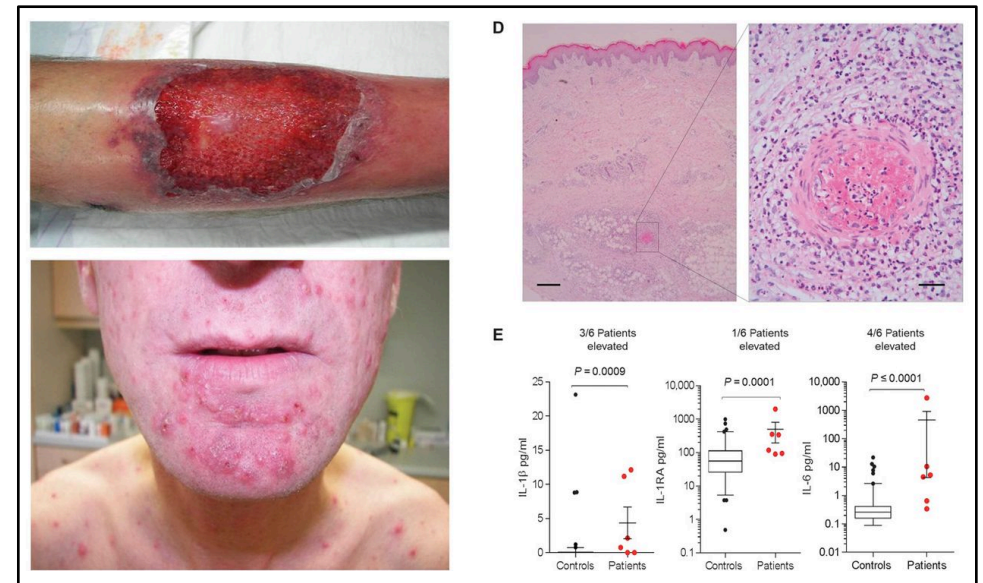


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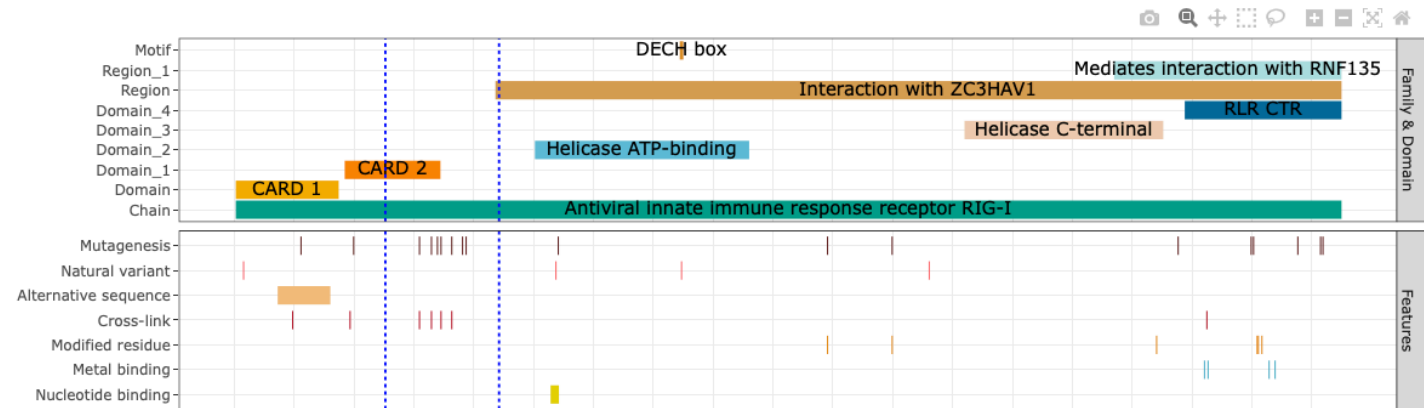


Protocol

Evidence source

Simple report

Search									
SYMBOL	Consequence	IMPACT	HGVSc	HGVSp	CANONIC AL	MAX_AF	MAX_AF_ POPS	gnomAD_exom es_non_topme d_POPMAX_A F	gnomAD_g mes_POPM _AF
DDX58	missense_variant	MODERATE	ENST00000379883.3:c.662N>G	ENSP00000369213.2:p.Asn221Ser	YES	0.0004539	AA	8.972790e-06	4.825090e-
DDX58	missense_variant	MODERATE	ENST00000379883.3:c.377N>G	ENSP00000369213.2:p.Asp126Gly	YES	0.0001462	gnomAD_A MR	1.467310e-04	2.939450e-

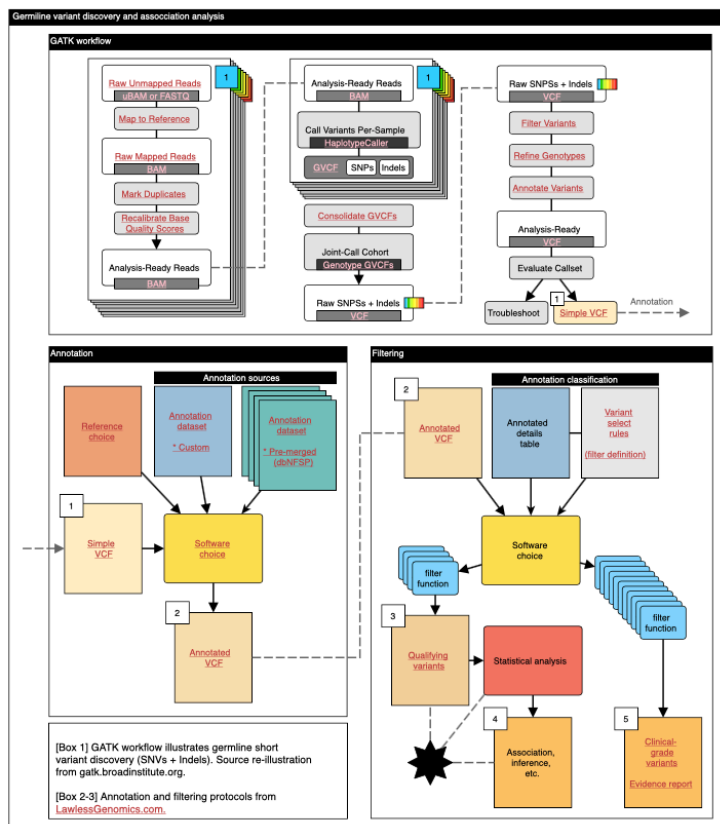


Protocol

Was the correct method used ?

How was it done ?

What does each step do ?



[Link: example protocol page](#)

Evidence source

Analysis browser.

Query report to access evidence sources.

Accurately report your finding

- ACMG standards and guidelines: for the interpretation of variants.
- Use accurate HGVS variant nomenclature: for precise clinical genetics.
- Use accurate HGNC gene names: for precise clinical genetics.
- Find other genes: outside of IUIS IEI genes.
- Coming soon: summarise all diagnostic evidence sources.

IUIS IEI table

Disease	Gene symbol	Inheritance	Inheritance detail	T cell count	B cell count	Immunoglobulin levels
CD3d deficiency	CD3D	AR		Very low	Normal	Low
CD3e deficiency	CD3E	AR		Very low	Normal	Low
CD3z deficiency	CD3Z	AR		Very low	Normal	Low
Coronin-1A deficiency	CORO1A	AR		Very low	Normal	Low
gc deficiency (common gamma chain SCID, CD132 deficiency)	IL2RG	XL		Very low	Normal to high	Low
IL7Ra deficiency	IL7R	AR		Very low	Normal to high	Low

PharmVar

Allele	Protein	Archival change (2010)	Transcript	Effect	Enzyme activity	Reference
CYP2A6*1	CYP2A6	None	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*2	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*3	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*4	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*5	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*6	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*7	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*8	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*9	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010
CYP2A6*10	CYP2A6	Y100H	AB0249	Normal	Normal	Shimada et al., 2010

[Link: example evidence page](#)

HGNC

Symbol report for IL7R

IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

Gene structure: IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

Gene expression: IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

OMIM

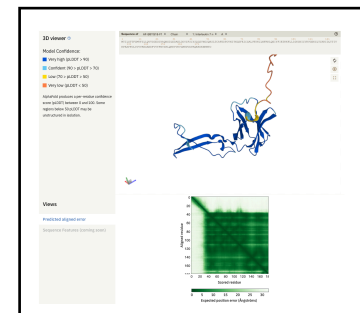
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Description: IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

Genetics: IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

Alpha fold



UniProt

UniProtKB S54 results

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Protein entry: IL7R (Interleukin 7 receptor) is a protein-coding gene located on chromosome 2 (2p11.2). It is a member of the IL7R family and is involved in the regulation of T cell development and function.

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GnomAD



ClinGen

Gene-Disease Validity

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many more

When you have an efficient tool
you open new doors

Clinical diagnosis is a
simple application example

Every step

- Drug / device development and regulation
- Genomic-based discovery
- Genomic medicine

Every field

- Pharmaceutical industry
- Personalised medicine
- Biomedical sciences

