AI in genomic medicine and rare disease research: a caregiver’s perspective

Roundtable on Genomics and Precision Health
October 2025

Will Greene
Rare Disease Researcher and Advocate
Board Member, Foundation for Prader-Willi Research

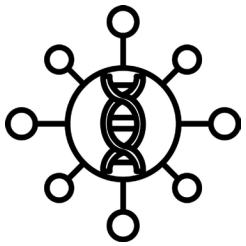
Disclosures & Background

No conflicts of interest but take my perspectives with a grain of salt anyway

No financial conflicts: I have no employer or income

Lead author on forthcoming paper with World Economic Forum building the case for greater investment in rare disease data

Recently employed as Healthcare Engagement Lead at Roche Diagnostics Asia Pacific (2020 – 2024)



Previously ran consultancy doing research, marketing, and business services for healthcare firms in Asia Pacific → project work in genomics, precision medicine, and AI topics

How AI powers my personal and professional life

I use it almost every day to support my son’s care and research/advocacy work

Care

Research

Advocacy

Our caregiver journey predates the current AI boom

It all began with Ari’s birth in December 2021

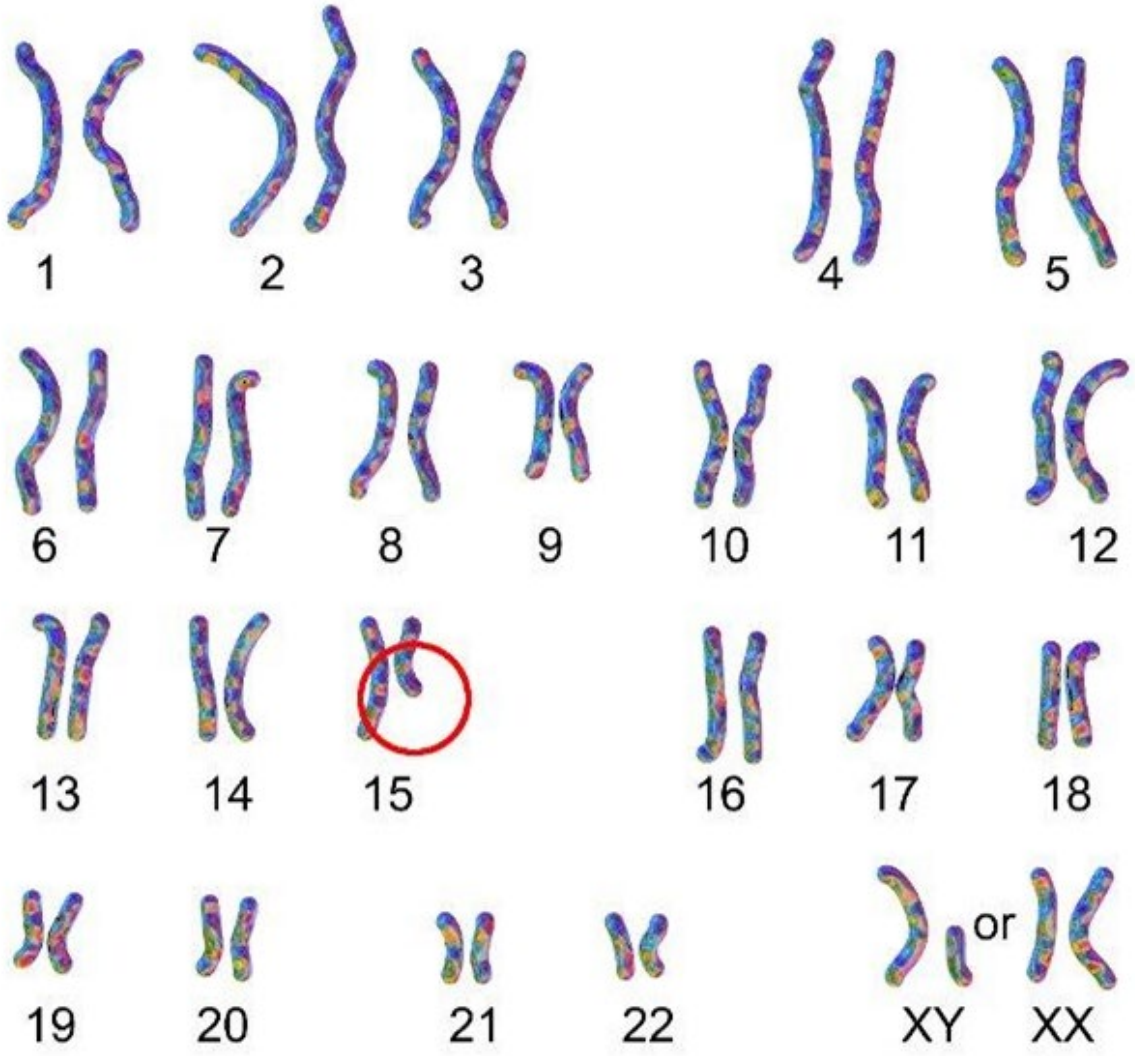
We knew right away that something wasn’t right

Ari had many of the signs and symptoms of a genetic condition

- Low birth weight
- Inability to cry or drink
- Hypotonia (floppy muscles)
- Other worrying signs

What is Prader Willi syndrome?

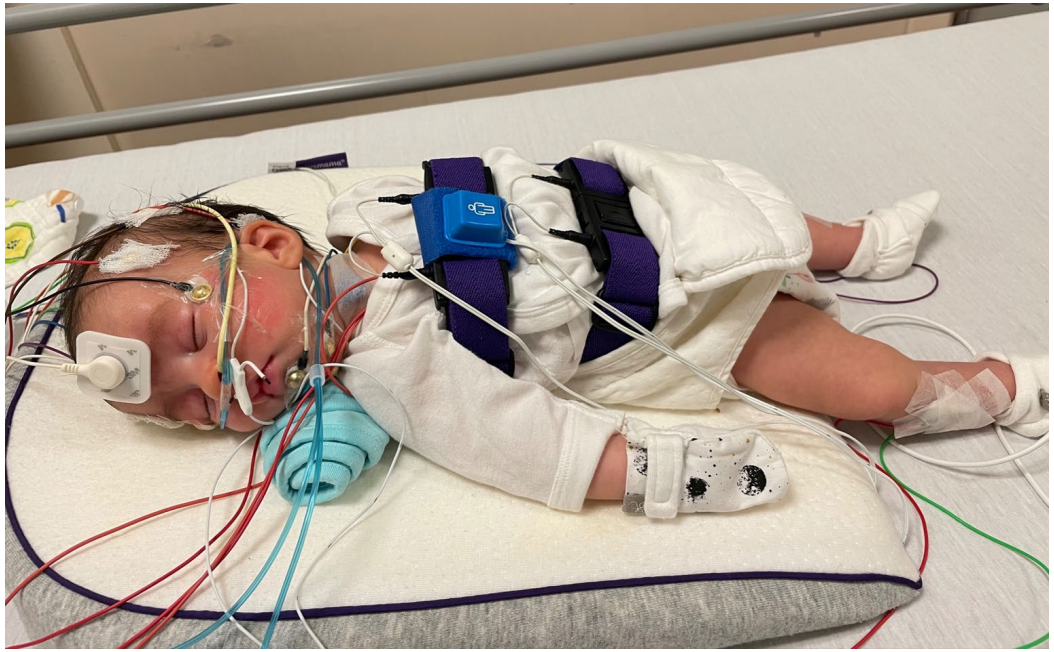
A rare and serious genetic disease impacting 1 in 15,000 live births



- Hyperphagia, i.e. feeling hungry all the time
- Anxiety and behavioral challenges
- Intellectual disability
- Developmental delays
- Other medical problems

No time for self-pity

We needed to get Ari on a treatment plan, and fast



First steps into rare disease research and advocacy

Raising awareness for PWS and rare disease research

FOUNDATION FOR PRADER-WILLI RESEARCH

Working Toward an Independent Future

Caring for an infant with PWS: the first few months

MEDIUM.COM

When our son Ari was diagnosed in late January with Prader-Willi Syndrome (PWS), a rare and serious genetic disorder, my partner and I began a ...

BabySeq, ICoNS and the power of newborn sequencing: Q&A with Dr Robert Green

Robert Green, Will Greene

Dr Phil Reilly on newborn screening: past, present and future

Phil Reilly, Will Greene 13 Oct 2022

Discovering AI after moving to the US

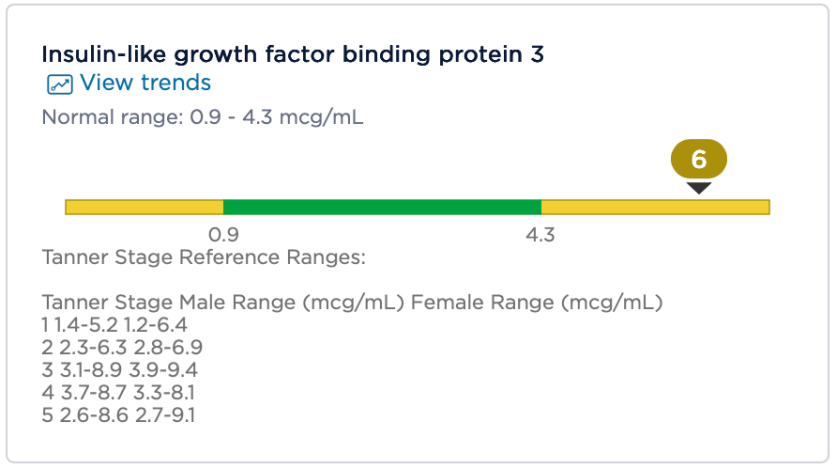
First in the context of reviewing grants for FPWR…oops

Advocate Reviewer Guidelines

Advocate reviewers will be selected based on prior experience in advocacy and general knowledge of PWS. They will evaluate the relevance of the proposal to the concerns of the PWS community. The advocates will evaluate the goals of the proposal with respect to the mission of the Foundation for Prader-Willi Research: ***to eliminate the challenges of PWS through the advancement of research.*** Reviewers are expected to maintain **strict confidentiality** regarding the applications, and use of AI, Large Language Models, or similar tools (like Chat GPT) for the advocate reviews are **strictly forbidden**. If you would like to become an advocate reviewer, please contact [redacted]@fpwr.org.

Using AI to manage our son's care

Privacy and data security are not particularly high concerns for us



- Note: The Kaiser system specifies that the "normal" range for IGF-I is 30-155 and IGFBP-3 is 0.9-4.3, based on the Tanner Stage Male Range for IGFBP3
- 2024
  - Oct 21 - shifted to Omnitrope
  - Nov 14 - Nov 14 - T4 and TSH in normal range; IGF-BP3 outside of normal range at 6.3 mcg/mL; IGF-1 outside of normal range at 177, same for ILGF-1 Z-Score at 2.8
    - Nov 24 - Gassner says move dosing down to 0.5mg/day and repeat labs 6 weeks after that
    - Nov 25 - Dr Stafford concurs, saying she sometimes lets IGF-1 to be above normal if IGF-BP3 is also high, as this would give a normal "free IGF-1," but because the Z-score is high (>2.5) and Ari is younger, she'd decrease too
  - Dec 4 - picked up GH stock
  - Dec 30 - new needle stock delivered
- 2025
  - Jan 6 - IGFBP-3 at 5.3 mcg/ML, slightly outside normal range; IGF-1 in normal range at 115 ng/mL; ILGF-1 Z-Score = 1.1.
    - Jan 13 - Gassner: I would recommend actually that we back off on dosing because the IGFBP3 is high, above range. The IGF1 is actually more a nutritionally dependent hormone and not as reliable as the IGFBP3 which is now above normal. So I would suggest a dose of 0.4 mg once/day, and we can repeat in 3 months the levels.
    - Jan 16 - Stafford: Gassner is correct that IGF-1 is more nutritionally dependent, but this is only generally applied to those who are nutritionally deprived and therefore have low IGF-1 and normal IGFBP-3. This assessment is used when looking at those with poor growth and investigating for growth hormone deficiency. I have never used this as an assessment of growth hormone dosing. I would not reduce the GH dose

Date	IGF-1 (ng/mL)	Z-Score	IGFBP-3 (mcg/mL)
Nov 14, 2024	177 ❗	2.8 ❗	6.3 ❗
Jan 6, 2025	115 ✅	1.1 ✅	5.3 ❗
Apr 15, 2025	120 ✅	1.3 ✅	4.7 ▲
Aug 6, 2025	220 ❗	3.8 ❗	6.0 ❗

Legend:

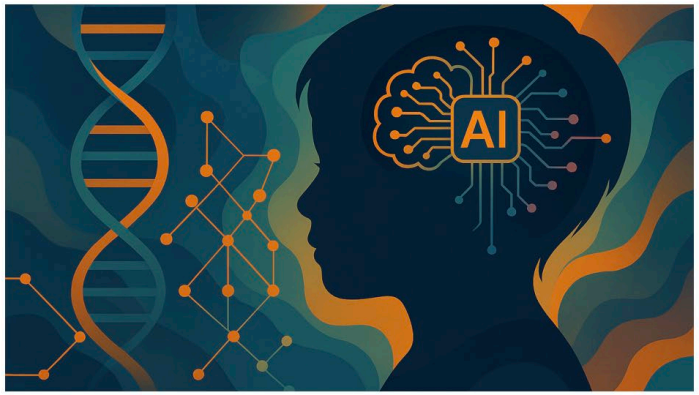
✅ = Within normal range

▲ = Near upper limit

❗ = Above range

Using AI for rare disease research and advocacy

I barely even write an email these days without running it through an AI



Three Ways AI is Changing Pediatric Genomic Medicine

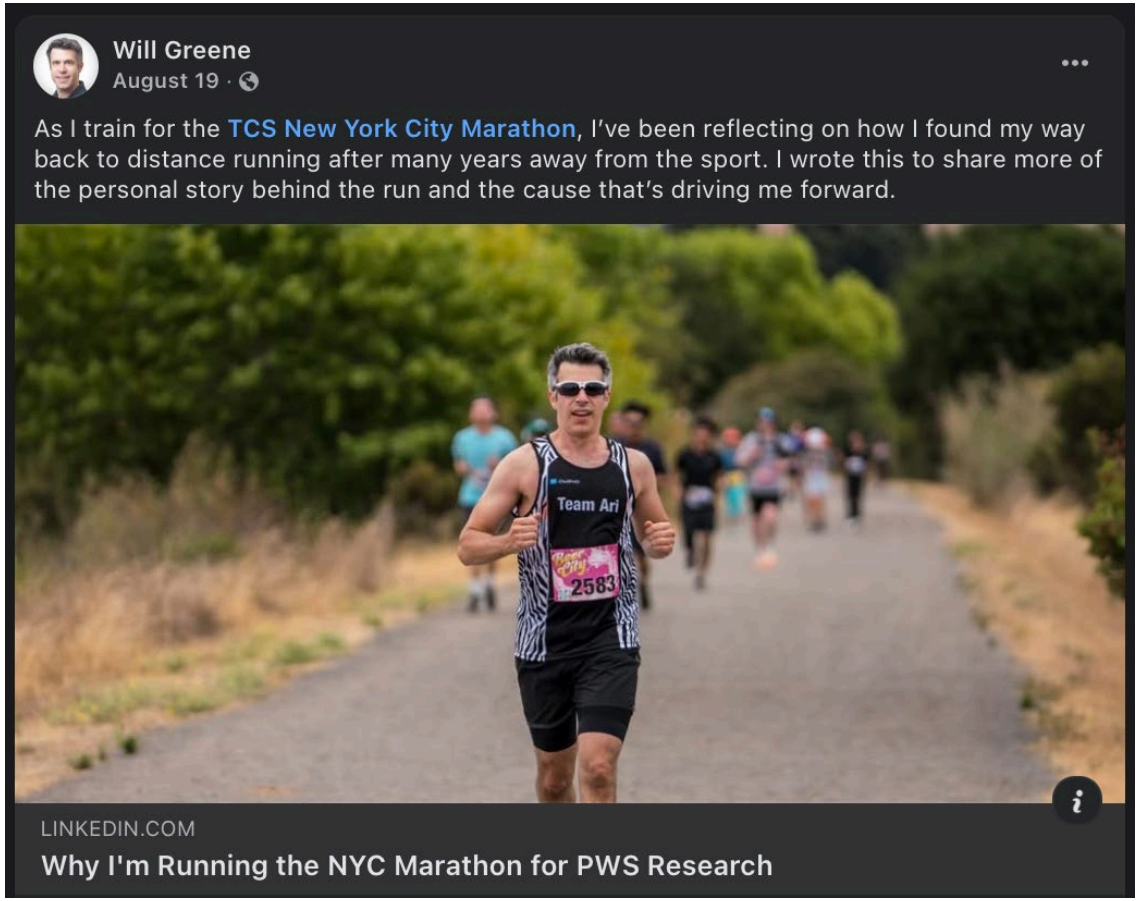
Will Greene
Rare Disease Research, Advocacy & Innovation

August 7, 2025

In early 2023, a year after my son was diagnosed with Prader-Willi syndrome (PWS), I began experimenting with newly emerging AI tools to support our family's care journey. They showed promise but offered little practical help at the time.

That changed dramatically last year, when large language models (LLMs) and other AI services became significantly more powerful. I now use them regularly as a caregiver, advocate, and researcher in the rare disease space.

Going forward, virtually every stakeholder in pediatric genomic medicine will need to grapple with these tools. AI is already becoming embedded into the way we diagnose, treat, and support children with rare genetic diseases. Its influence will only deepen in the years ahead.



Final thoughts

AI is still imperfect but pretty darn useful

- Patients and caregivers are using AI for everything, and unlikely to stop
- Data privacy matters, but for many caregivers, quick answers matter more
- We still need human intelligence to point AI in the right direction