Rare Disease: Busting Myths, Fueling Miracles

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Rare Diseases: World's Largest Invisible Medical Population

350M

People Affected

7K

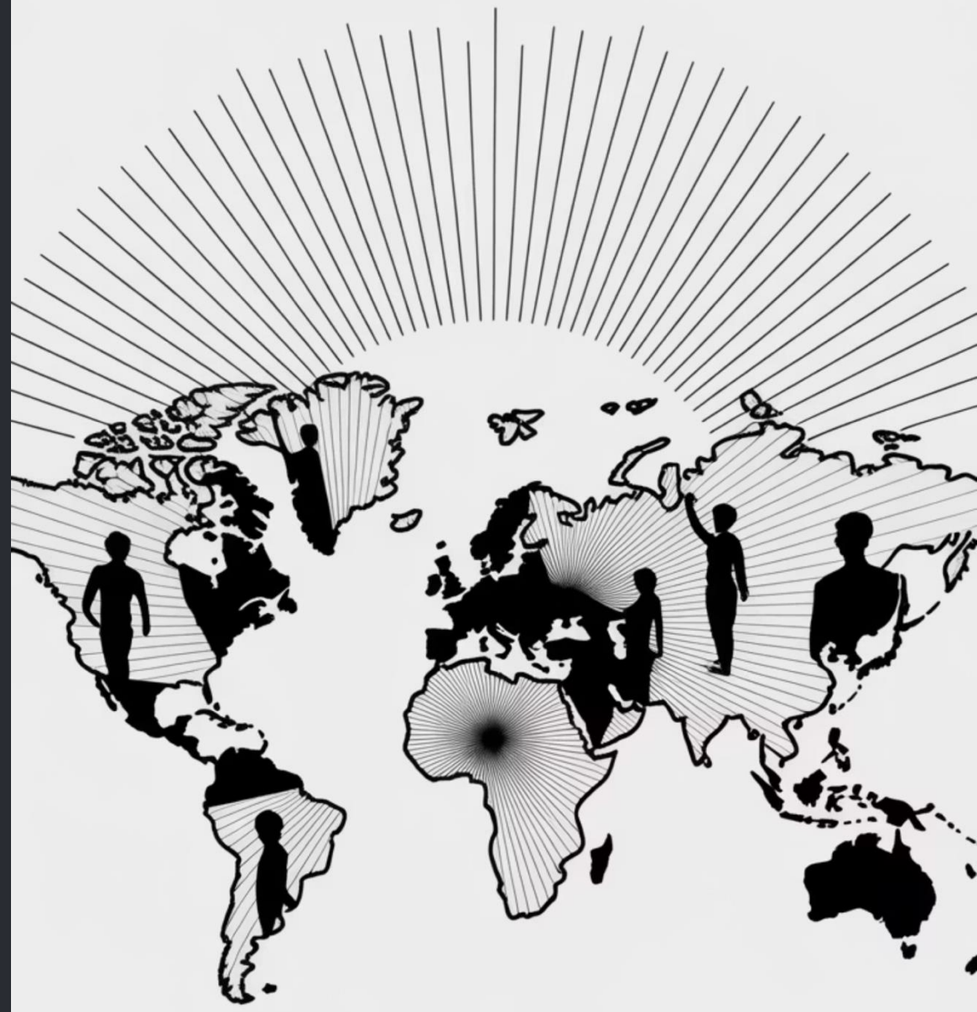
Conditions

95%

Untreated

Every accurate story illuminates another dark corner of the map.

Your pen is light.



FONDATION IPSEN

Our Mission for People Living with Rare Disease

Fondation Ipsen, an independent global philanthropic organization, addresses these challenges through education, advocacy, and research.

EDUCATE

ADVOCATE

CURE

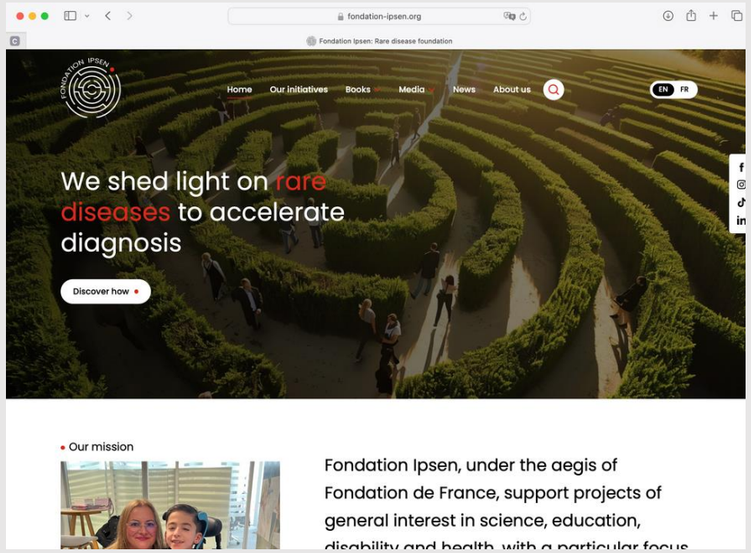
Fondation Ipsen

989,000
Books

10.5M
Digital Reach*

282
Global Experts

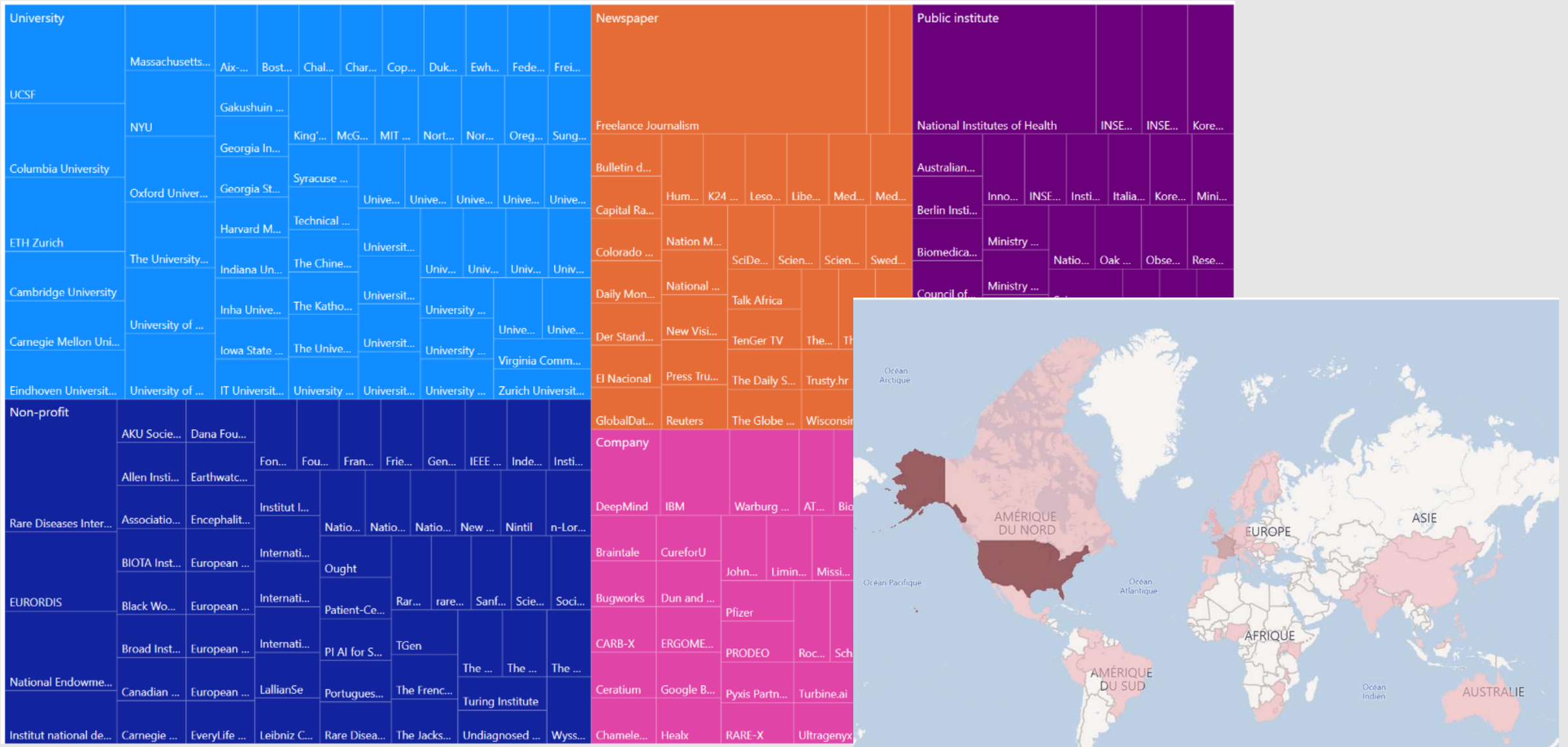
120+ countries.



*Primary reach only**

240 organisations in 50 countries.

Powered by scientific experts: 282 independent experts



Fondation Ipsen's global expertise map

Science backbone

Scientific needs assessments
45 rare disease associations

- Scientific methodology
- Peer reviewed
- Published

ELSEVIER

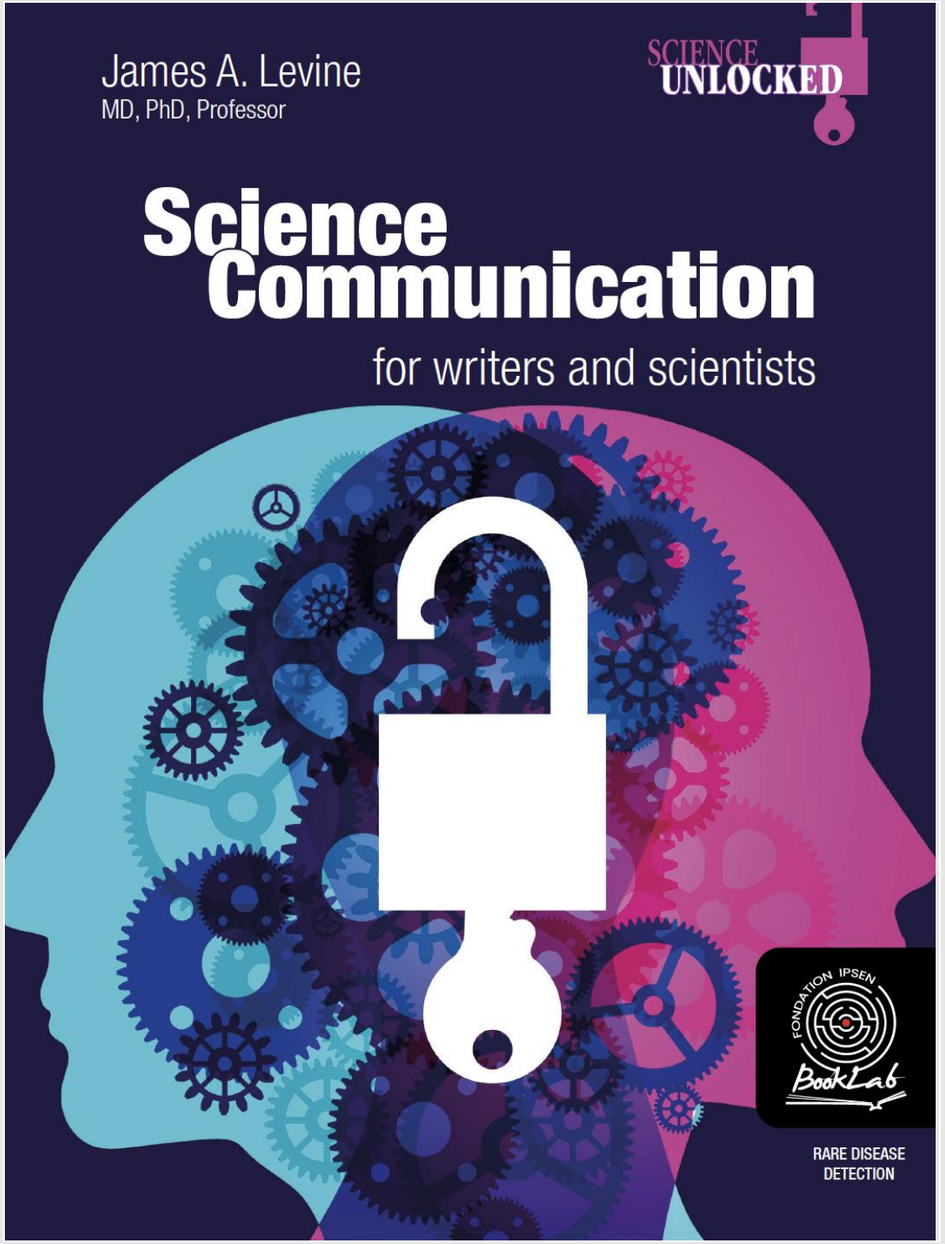
Rare

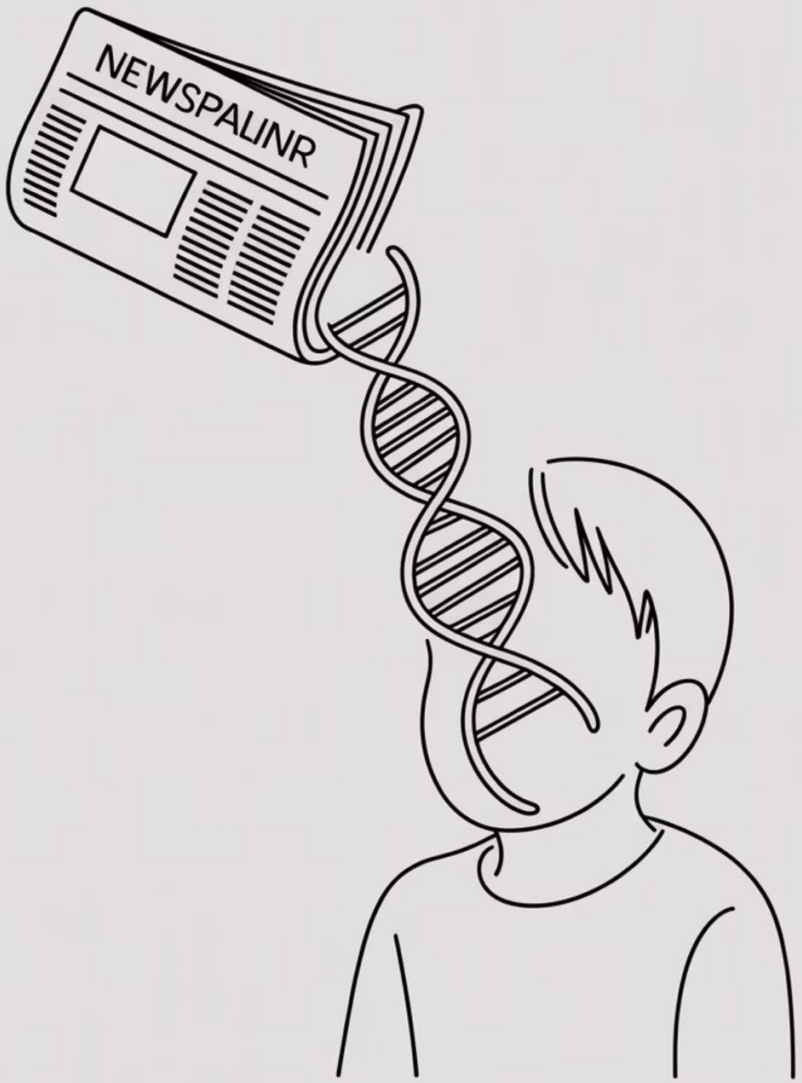
Volume 3, 2025, 100064

A qualitative needs assessment of external communication by rare disease associations.

J.A. Levine [a], S. SiWan Zimmerman [b], F. Delval [a], L. Perkins Smith [b], A. Kitchen [b], E. Garrigues Tena [a]

https://www.amazon.com/Science-Communication-writers-scientists-Briefings-ebook/dp/B0FG39N866





A Child Finally Has a Name

One headline can outlive a thousand papers.

A family emerging from years of uncertainty.

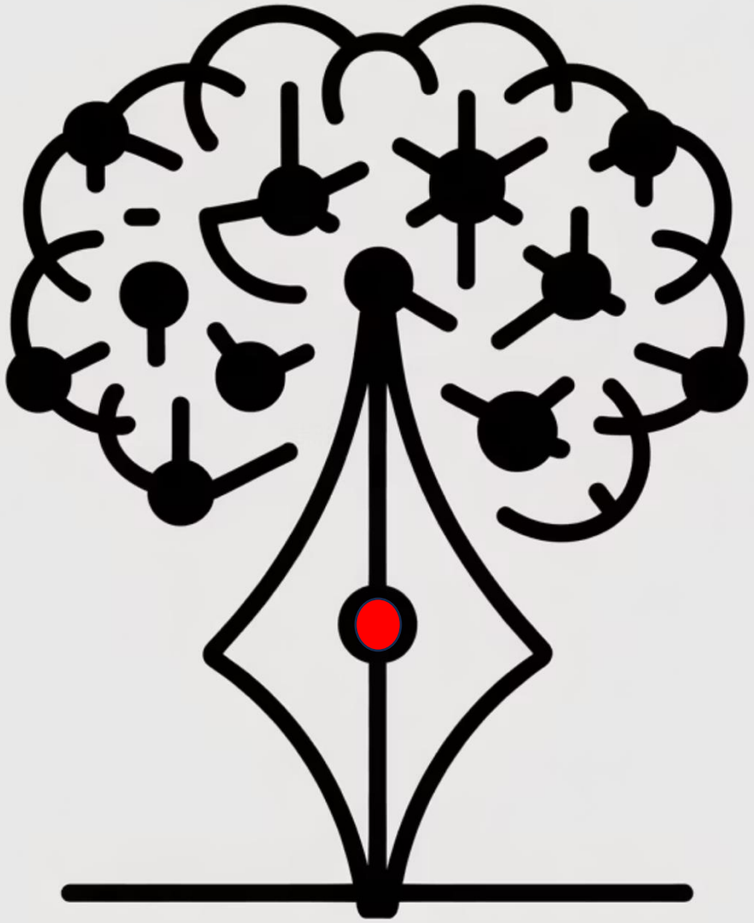
Words can be diagnostic tools = oxygen.

Words, when placed with care, heal.

Journalism
– the Oxygen of Science

Without storytellers, discoveries **suffocate in jargon**. You translate research into relevance and empathy.

Translators between two cultures:
the language of molecules and the language of meaning.



Innovation Communication Paradox

We Can Sequence a Genome...
But Not Tell Its Story

Technology races; narrative lags.

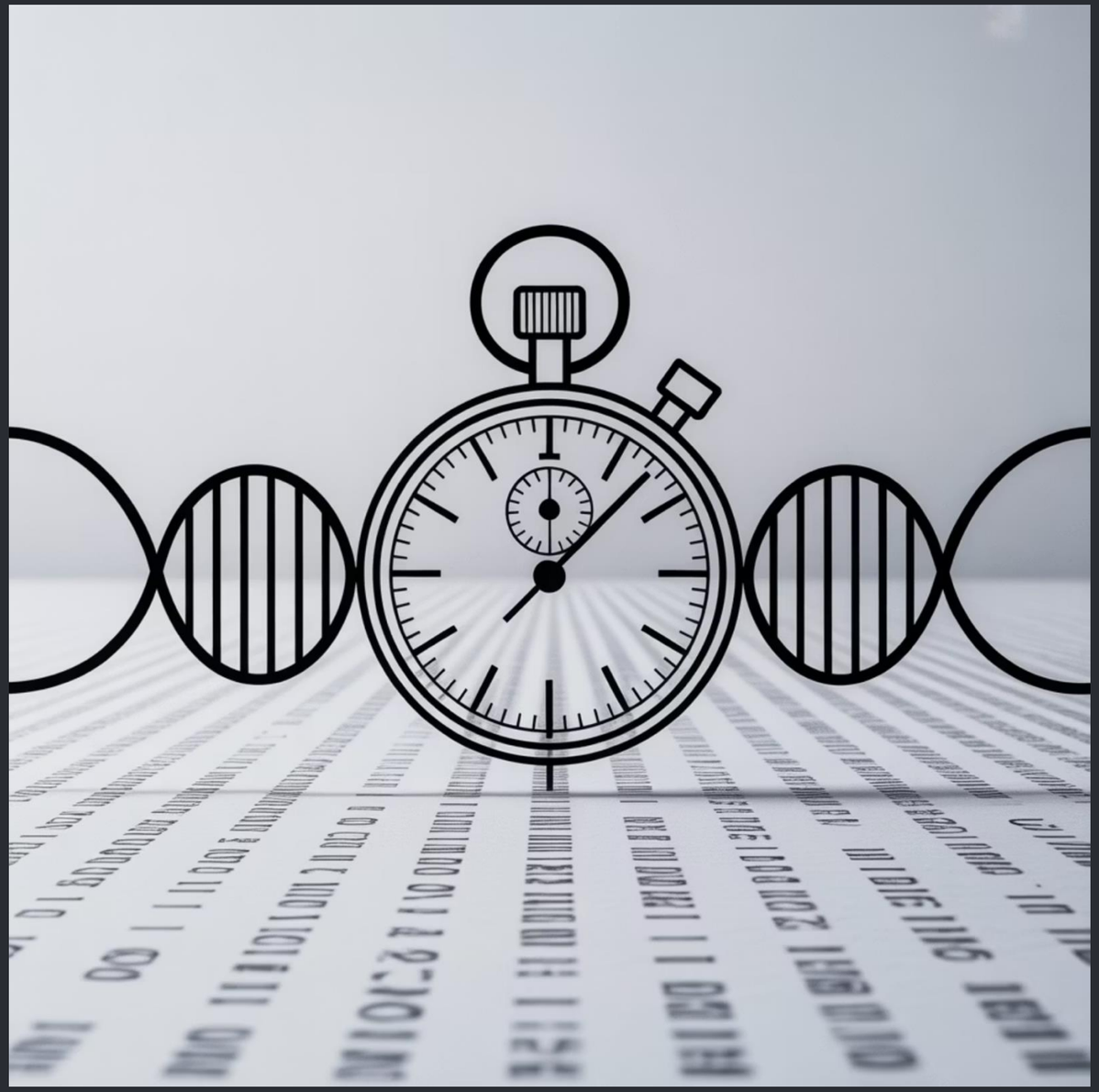
We can decode a genome overnight

but struggle to explain what that means for a family.

CRISPR dominates headlines.

Innovation without explanation is invention

without trust.



From Disease to Design

The New Language of Health

Medicine is becoming creative engineering.

The lexicon shifts from cure to construction, patient to partner.

Journalists can teach society this new grammar.

Every story you tell expands the public vocabulary of progress.

Empathy: a Form of Evidence

1

Data Convinces

Facts change minds with precision and proof

2

Feeling Commits

Emotion changes behavior and drives action

Compassion boosts recall.

Write with precision—and pulse.

Heart plus head.

The Scientist, the Parent, the Algorithm

The Scientist

Rigorous research and clinical expertise

The Parent

Lived experience and urgent advocacy

The Algorithm

Data analysis and pattern recognition

Three voices together.

Challenge: orchestrate their harmony.

How to Be Awe-Struck Without Being Fooled

The Ethics of Wonder

Hype

Exaggerated claims erode credibility

Hope

Realistic optimism grounded in evidence

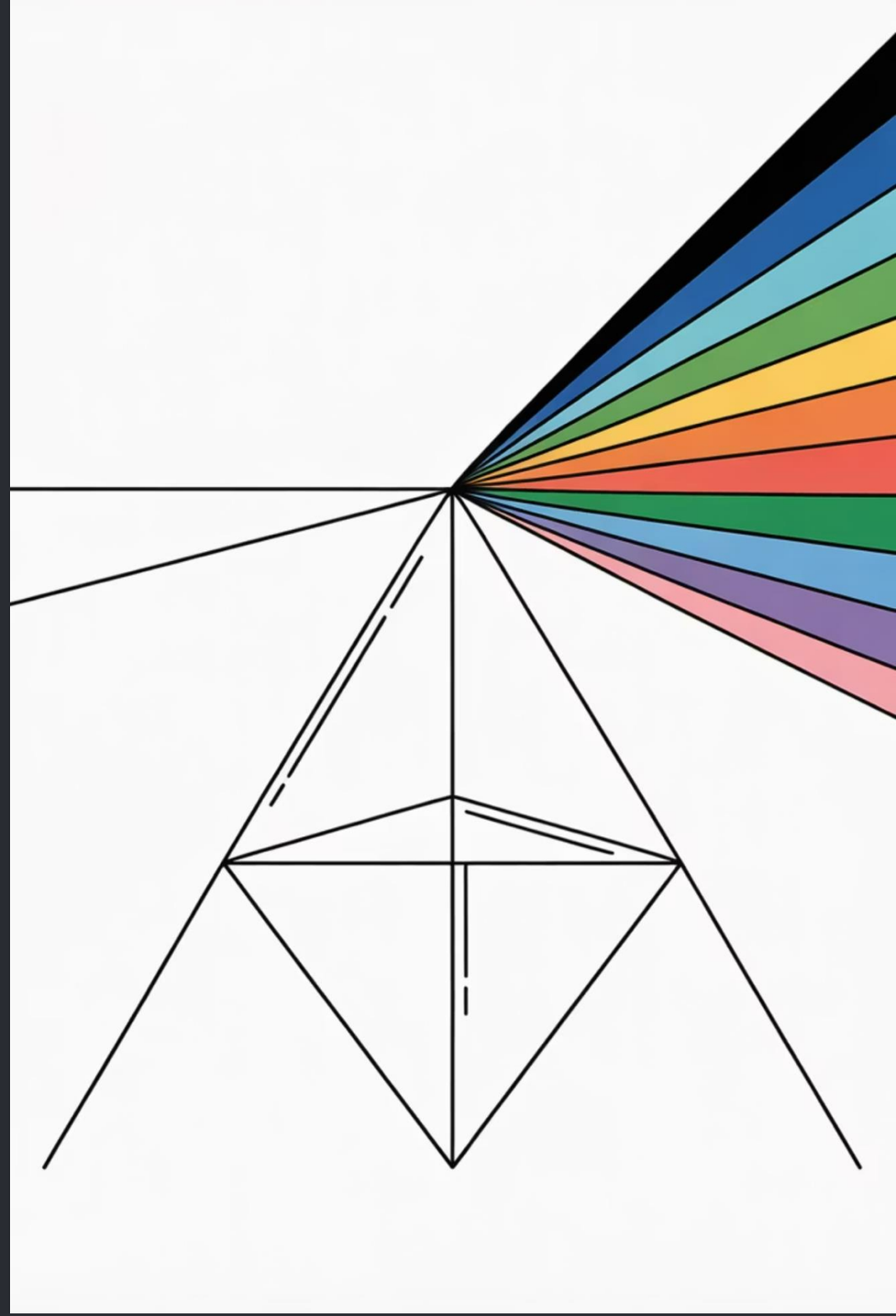
Honesty

Transparent reporting of limitations

The fault line between awe and accuracy.

"Breakthrough" headlines risk hype.

Curiosity + skepticism = credibility.



From Victims to Visionaries

Breaking the Pity Narrative

People with rare diseases are NOT waiting to be saved.

They are building the future themselves: researchers, advocates, entrepreneurs, and innovators.




Agency, not tragedy. Protagonists, not footnotes.

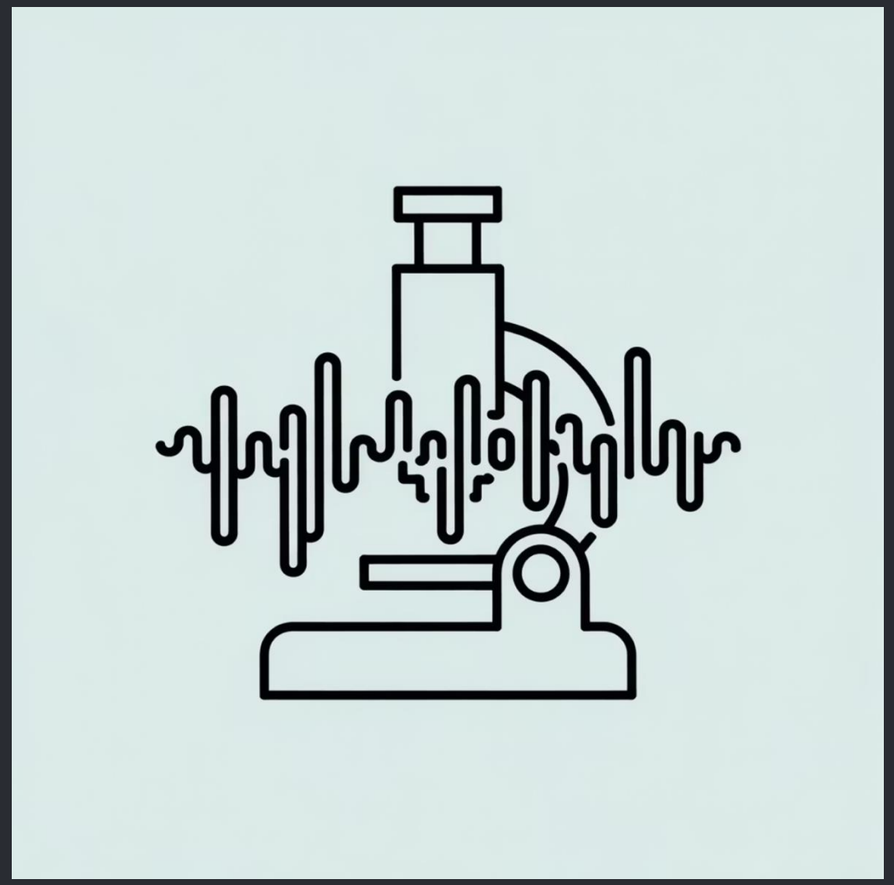
Language shapes perception, and perception shapes possibility.

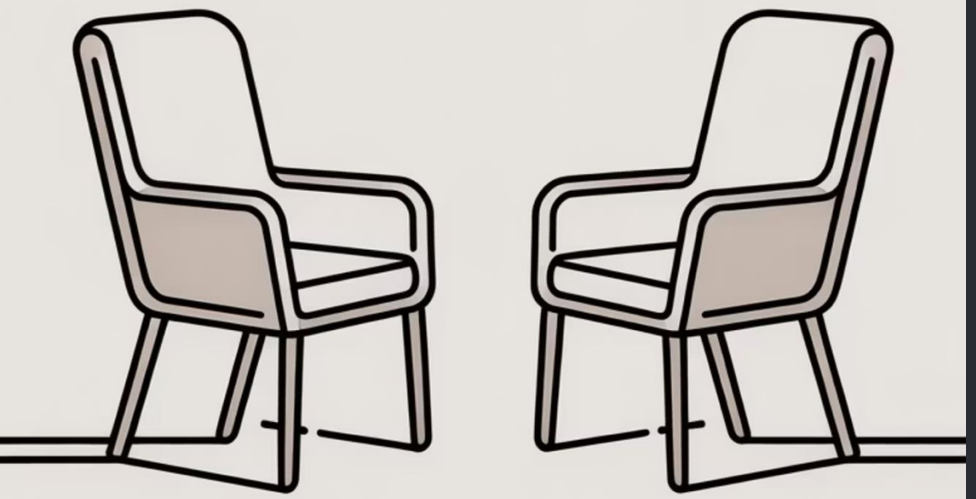
The Scientist Interview

"What Keeps You Awake at Night?"

Every discovery hides emotion: doubt, obsession, relief, fear, and triumph.

- Ask about failures, not just successes
- Explore the personal cost of persistence
- Find the moment of uncertainty before breakthrough
- Connect the science to the scientist's story





The Patient Interview

"What Do You Want the World to Know?"

01

Move from extraction to collaboration

Let patients co-author their story

02

Dignity alongside accuracy

Accuracy and dignity are inseparable

03

Agency over adversity

Focus on building, not just enduring

Progress—Not Perfection

The Grammar of Hope

Afiyah Raven, Learner

The DACMAR Lens

Six Questions for Every Innovation Story

Disruption
What changes?

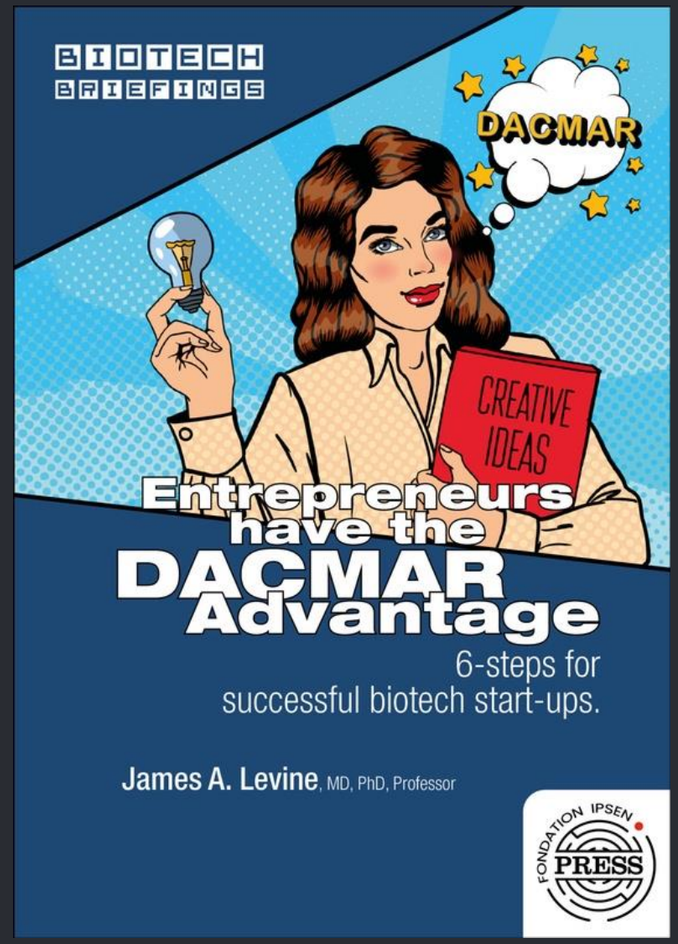
Adoption
Who uses it?

Resources
What does it cost?

Collaboration
Who partners?

Adaptability
Can it scale?

Management
How is it governed?



DACMAR is curiosity with guardrails—a framework for comprehensive, responsible reporting.

Thank you

Myths about Rare Disease

Key Issues in Rare Disease

10 Myths about Rare Disease

Myth #1: “Rare diseases affect almost no one.”

The Reality

- 300 million people live with a rare disease — equivalent to the population of the U.S.
- Collectively, rare is common; nearly every family knows someone affected.
- One in 17 people will experience a rare disease at some point in life.
- The misconception minimises urgency, funding, and political attention.

Why It Matters

Scale shapes strategy. Rare disease is a global public-health priority, not a niche category.

Myth #2: “If a disease is genetic, nothing can be done.”

The Reality

- Genetic does **not** mean untreatable.
- Gene therapy, ASOs, RNA editing, and small molecules now target genetic roots.
- Early diagnosis enables proactive care, slowing or preventing complications.
- Supportive therapies—respiratory care, nutrition, physio—dramatically improve survival and quality of life.

Why It Matters

Genetics is a starting point, not a dead end

Myth #3: “Rare diseases always appear at birth.”

The Reality

- Many rare diseases are **adult-onset**: ALS, Huntington’s, many metabolic and immune disorders.
- Symptoms may be triggered later by environment, hormones, stress, or infections.
- Adults often face misdiagnosis because rare conditions are viewed as “paediatric problems.”

Why It Matters

Awareness across the lifespan prevents decades of delayed care

Myth #4: “Diagnosis is impossible without obvious physical symptoms.”

The Reality

- Many rare diseases present with subtle, fluctuating, or invisible symptoms.
- Pain, fatigue, neurological issues, and behavioural changes are often misunderstood.
- Advanced diagnostics (genomics, metabolomics, AI-assisted imaging) can reveal hidden pathology.
- Lack of visible signs leads to stigma and dismissal.

Why It Matters

Invisible does not mean imaginary. Precise tools + awareness = faster answers.

Myth #5: “There are no treatments for rare diseases.”

The Reality

- There are **hundreds** of approved orphan drugs, gene therapies, ASOs, and enzyme replacements.
- Global pipelines are expanding rapidly—over 1,000 therapies in active development.
- Many symptoms can be managed effectively even without disease-specific drugs.
- Access, not availability, is often the barrier.

Why It Matters

The landscape is changing fast: treatment deserts are not inevitable.

HOWEVER: NO CURES FOR 94% OF PATIENTS LIVING WIRTH RARE DISEASES

Myth #6: “Developing therapies isn’t worth it for small populations.”

The Reality

- Venture philanthropy has proven otherwise (CF Foundation → Vertex).
- Precision medicine lowers R&D risk by targeting well-defined biology.
- Regulators offer strong incentives: ODD, PRIME, ILAP, RMAT.
- Platform technologies (AAV, CRISPR, ASO) spread cost across multiple diseases.

Why It Matters

Small numbers do not equal small impact. Rare disease often pioneers the science later adopted for common diseases.

Myth #7: “Patients with the same rare disease are all alike.”

The Reality

- Phenotypes vary widely even within a single gene mutation.
- Disease progression differs by age, modifier genes, environment, and comorbidities.
- “One-size-fits-all” treatments and guidelines often fail.
- True precision medicine requires personalised monitoring and care plans.

Why It Matters

Variation is the rule, not the exception. Care must reflect individuality.

Myth #8: “Rare disease care ends once a diagnosis is given.”

The Reality

- A diagnosis is **not the finish line**—it’s the start of lifelong navigation through medical, social, educational, and psychological systems.
- Many patients still lack coordinated care, treatment plans, or specialist follow-up after receiving a name.
- Families face ongoing challenges: managing symptoms, accessing therapies, dealing with emergencies, updating care plans, and navigating transitions (school → adulthood → ageing).
- Adult care is especially fragile: most systems are built for paediatrics, not lifelong needs.
- Without continuous support, patients fall through the cracks—even with a confirmed diagnosis.

Why It Matters

A name opens the door, but it doesn’t build the pathway. Rare disease requires lifelong care, not a moment of recognition.

Myth #9: “Technology will solve rare disease automatically.”

The Reality

- AI, genomics, and data platforms accelerate progress—but require governance, trust, and interpretation.
- Algorithms cannot replace lived experience or clinical judgement.
- Digital divides leave many communities behind.
- Technology amplifies inequities unless deployed intentionally.

Why It Matters

Tech is a tool, not a solution. Human expertise and patient partnership remain central.

Myth #10: “Patient organisations are small charities with limited influence.”

The Reality

- Patients drive research, set agendas, create natural-history studies, and fund clinical trials.
- They negotiate with regulators, shape reimbursement policy, and influence pharma strategy.
- Venture-philanthropy funds now catalyse billion-dollar transformations.
- Many breakthroughs began with a kitchen-table parent group.

Why It Matters

Patients are not beneficiaries—they are leaders, innovators, and investors.

10 Key Issues in Rare Disease: Levers of hope

1 — The Diagnosis Odyssey

The Challenge of Getting a Name

7–10 years to diagnosis on average; many never receive one.

- Fragmented pathways: GP → specialist → tests → more specialists → still no clarity.
- Symptoms often rare, subtle, or mimic common childhood or adult conditions.
- Limited clinician awareness: most physicians will never see more than one case.
- Genetic testing exists, but access, cost, and interpretation remain uneven.
- Families experience grief, self-doubt, and profound uncertainty throughout.

Why It Matters

A delayed name delays treatment, support, school accommodations, and hope.
Diagnosis is not an endpoint—it’s the beginning of understanding.

2 — The Therapeutic Desert

95% of Rare Diseases Have No Approved Treatment

- Biology is complex, heterogeneous, and poorly mapped.
- Pharma incentives remain limited despite progress in the Orphan Drug Act.
- Small populations → high R&D risk → low commercial return.
- Clinical endpoints unclear; natural history often undocumented.
- Diseases sometimes progress too fast—or too unpredictably—for standard trial design.

Why It Matters

Innovation stalls in the absence of clear commercial pathways. Patients wait while science advances but translation lags.

3 — Fragmented Healthcare Pathways

A System Not Designed for the Exceptions

- Specialists rarely coordinate; communication breaks across departments.
- Emergency care teams often unaware of condition-specific needs.
- Transition from paediatric to adult care is a cliff edge.
- Rural and underserved communities face even larger barriers.
- Paperwork overload: disability benefits, rare disease exemptions, school plans.

Why It Matters

Fragmentation turns complexity into suffering. A condition may be rare—but complexity is universal.

4 — Inequity in Access

Where You Live Determines the Care You Receive

- Diagnostic tests available in Paris or London may be inaccessible in rural India or sub-Saharan Africa.
- Even within wealthy regions, disparities persist across socioeconomic lines.
- Access to clinical trials overwhelmingly favours big cities.
- Cost of orphan drugs can exceed **€200k–€700k per year**.
- Reimbursement policies vary dramatically across countries.

Why It Matters

Geography should not be destiny. Yet for millions, it is.

5 — Data Without Governance

The Rare Disease Data Paradox

- Rare disease science depends on pooling small datasets.
- Registries are uneven, fragmented, and often incompatible.
- Concerns over ownership, commercial use, and patient trust.
- AI tools learn best from large, well-curated datasets—but rare disease data is often sparse or siloed.
- Families fear misuse of genetic information.

Why It Matters

Data is the new lifeline—but only if trust and governance exist.

6 — Burden on Families and Caregivers

The Hidden Workforce of Rare Disease

- Parents become care coordinators, nurses, advocates, and exhaustively informed experts.
- Caregiving hours regularly exceed **40–60 hours per week**.
- Siblings often feel unseen or displaced.
- Financial strain: reduced employment, travel to specialists, uncovered therapies.
- Psychological toll: chronic stress, social isolation, anticipatory grief.

Why It Matters

Families are the real infrastructure of rare disease. Supporting them is not optional —> it is central.

7 — The Clinical Research Bottleneck

Trials Are Not Built for Small Populations

- Recruiting 15 patients can take 3 years.
- Traditional RCTs often impossible or unethical.
- Outcomes vary widely across individuals—statistical power evaporates.
- Natural history studies take years to establish baselines.
- Regulatory pathways still catching up, despite ILAP, PRIME, ODD reforms.

Why It Matters

We have the science. What we lack are trial models fit for reality.

8 — The Silence Problem

Invisible Conditions Create Invisible People

- Many rare diseases have no patient organisation.
- Media coverage is episodic, tragedy-framed, or sensationalised.
- Awareness among policymakers is low; policy becomes reactive, not strategic.
- Stigma persists: misunderstood behaviours, physical differences, or uncertain prognoses.
- Lack of public language makes invisible suffering even quieter.

Why It Matters

What is not seen is not funded. And what is not funded does not improve.

9 — Transition and Lifelong Care

Children Survive Into Adulthood—But Systems Don’t

- Medicine improves; children who once died now live into adulthood.
- Adult physicians often lack rare disease experience.
- Adult social services, universities, employers rarely prepared.
- Gaps in mental health support during adolescence.
- Reproductive counselling, fertility, and adult independence are often overlooked.

Why It Matters

Rare disease is lifelong. Care should be too.

10 — Sustainability of Patient Organisations

The Fragile Backbone of the Rare Disease Ecosystem

- Most associations operate with **1–3 volunteer staff**.
- Funding unpredictable; burnout extremely common.
- Organisations are asked to be everything: advocates, educators, policy experts, research funders, conference organisers, mental-health providers.
- Digital transformation demands skills and tools most associations simply don’t have.
- Venture philanthropy outperforms, but adoption is uneven and often misunderstood.

Why It Matters

A strong rare-disease ecosystem needs strong foundations. Without sustainable patient organisations, nothing truly scales.

Awareness to Impact: Journalism the Catalyst

You are the oxygen for change

Journalism can report facts, or it can illuminate pathways for change. The words we choose determine whether readers remain passive observers or become active participants in solving complex challenges.

Every headline is an opportunity to mobilize for impact.