A PETITION FROM THE RARE AND ORPHAN DISEASE COMMUNITY OF INDIA

The Uncounted: Why India Must Find, Recognize, and Care for Its Rare Disease Patients

Rare diseases are rare only one at a time. Together, more than 7,000 of them affect an estimated one in every ten to twenty people. In India that is tens of millions of citizens, most of them children, and most of them have never been diagnosed. This is a plea to count them, because a country cannot care for people it refuses to see.

Submitted to the Government of India for the attention of the Union Ministry of Health and Family Welfare, the Department of Health Research, and the Governments of the States. Prepared with, and to be co-signed by, India's rare-disease patient organizations, physicians, medical geneticists, and international partners.

Introduction: a problem hidden by its own definition

A disease is called "rare" when few people have it. That single word has done immense harm. It makes each condition sound like someone else's problem, a curiosity at the far edge of medicine. The truth is the opposite. There are so many rare diseases that, added together, they are common. The World Health Organization now recognizes that over 300 million people worldwide live with a rare disease, that there are more than 7,000 such diseases, and that about 70 percent of them begin in childhood.[1,2,3] Roughly 95 percent of these diseases still have no medicine that treats the disease itself.[4,8]

India has never measured how many of its own people are affected. Every number used for India is borrowed from studies done in the West.[3] That gap in knowledge is the single most important fact in this document, and it is the reason we write.

The problem, stated plainly

When a disease is not counted, three things follow, and all of them cost lives. First, doctors are not trained to recognize what they are told is vanishingly rare, so children are misdiagnosed for years. Second, governments cannot plan services, screening, or budgets for a population whose size is unknown. Third, families are left alone, often bankrupted, searching for a name for their child's suffering. A patient in India, like patients everywhere, can spend years and see many doctors before anyone identifies the illness.[9,10] For a child with a treatable disease, those lost years are the difference between a full life and an early death.

The current landscape in India

India has taken real first steps, and they deserve acknowledgment. The National Policy for Rare Diseases of 2021 created a framework, named 63 diseases for support, and set financial help of up to Rs 50 lakh per patient.[11] The number of government Centres of Excellence has grown from 8 to 15, now including AIIMS Patna in Bihar.[12] India has built a national registry that has enrolled 15,369 patients.[14]

And yet the system reaches almost no one. In the policy's first years, only 1,118 patients received treatment support under it.[13] In the financial year 2025-26, of about Rs 299 crore set aside for rare-disease treatment, only about Rs 31 crore was spent, and roughly Rs 271 crore was set to lapse unspent.[15] The larger fund ordered by the Delhi High Court has been held up in the courts and is not yet reaching patients.[16] The money is not the first problem. The first problem is that we have not found the patients. Funds meant to save lives are going back to the treasury while children die waiting.

The projected numbers, and the objections we hear

Applying the globally accepted rate of 3.5 to 5.9 percent to our population gives roughly 70 to 100 million Indians living with a rare disease.[3] That number provokes disbelief, so here are the objections we hear, and our answers.

"It is impossible that 100 million Indians have rare diseases." It is the same rate the world lives with; the United States, which counts, finds 1 in 10.[4,5] A low Indian count measures how little we look, not how few are ill, and marriage within communities raises the odds of inherited disease here.

"America is different from India." The difference is that India does not screen or register its patients, so it does not see them. A cancer no one tests for is still cancer.

"India cannot handle common diseases, so why chase rare ones?" The tools that find rare diseases (newborn screening, genetic testing, registries) strengthen care for everyone, and many rare diseases are common illnesses misdiagnosed.

"If we find them, who treats them and who pays?" Most common rare diseases are treatable, several of them cheaply, and early diagnosis costs far less than the years of wrong tests and emergencies that follow a missed one.[4] A birth screen for phenylketonuria costs little and prevents a lifetime of disability.

"If they live longer, the population will grow." These are overwhelmingly children. A nation is measured by whether it protects its most vulnerable, not by whether it finds them inconvenient.

The gap, and what we ask

The deepest gap is between what we believe and what we know: we run a national policy on a number no one has measured. India already proved it can measure health at national scale with the ICMR-INDIAB study across 31 states and union territories.[19] It has never pointed that capability at rare diseases. We ask for three things.

One, count them. India's first population-based, multi-centre study of how many Indians live with rare diseases, across regions, languages, and incomes, with genetic confirmation and a registry that feeds and strengthens the ICMR national registry rather than building a parallel one.

Two, reach them. A national, AI-assisted virtual Centre of Excellence that extends the 15 physical centres to every district through the existing Ayushman Arogya Mandir primary-care network and the ABDM digital-health backbone, helping front-line doctors recognize warning signs, guiding families to the nearest specialist, and supporting caregivers in their own language. It supports doctors rather than replacing them, and it does not diagnose or prescribe on its own.

Three, spend what is committed. Fix the bottleneck that keeps funds from patients, which is that the patients have never been found.

> **The ask in one line.** Count India's rare-disease patients, then build recognition, support, and treatment that reach every district on India's existing rails (screening, digital, and registry infrastructure, and the National Policy for Rare Diseases treatment fund, not the Ayushman Bharat insurance cap), so that money already set aside stops lapsing while children die waiting.

References and sources

Every figure in the two-page summary is drawn from a government record, an inter-governmental body, or peer-reviewed research. Advocacy estimates are labeled as such. Sources were reviewed on 6 August 2026.

1. **World Health Organization.** Resolution WHA78.11, "Rare diseases," adopted by the 78th World Health Assembly, 24 May 2025. Recognizes over 300 million people living with more than 7,000 rare diseases, about 70 percent beginning in childhood, and mandates a Global Action Plan.
2. **United Nations General Assembly.** Resolution A/RES/76/132, "Addressing the challenges of persons living with a rare disease and their families," adopted 16 December 2021. Cites an estimated 300 million persons worldwide.
3. **Nguengang Wakap S, Lambert DM, Olry A, et al.** "Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database." European Journal of Human Genetics 2020;28(2):165-173. Point prevalence 3.5 to 5.9 percent; about 72 percent genetic; about 70 percent pediatric onset. (The peer-reviewed basis for the 300 million figure and for extrapolations to India.)
4. **Kaufmann P, Pariser AR, Austin C** (NIH National Center for Advancing Translational Sciences). "From scientific discovery to treatments for rare diseases." Orphanet Journal of Rare Diseases 2018;13:196. About 30 million people in the United States, or 1 in 10 Americans; fewer than 5 percent of rare diseases have an effective approved treatment.
5. **National Organization for Rare Disorders (NORD), USA.** "1 in 10 Americans" living with a rare disease; more than 25 million Americans. United States Orphan Drug Act, 1983 (defines a rare disease as affecting fewer than 200,000 people).
6. **EURORDIS-Rare Diseases Europe.** Approximately 30 million Europeans live with a rare disease; EURORDIS estimates that 6 to 8 percent of people are affected over a lifetime.
7. **World Health Organization, ICD-11** and Rare Diseases International. Operational description of rare disease (1 in 2,000 or fewer), Orphanet Journal of Rare Diseases 2024; ICD-11 codes approximately 5,500 rare diseases.
8. **Global Genes; The Lancet Global Health.** "The landscape for rare diseases in 2024," editorial, Lancet Global Health 2024. Restates that about 95 percent of rare diseases have no approved treatment, and cites the community estimate that about 30 percent of children with a rare disease die before age 5 (an advocacy-origin estimate, presented here as such).
9. **Faye F, Crocione C, Anido de Pena R, et al.** (EURORDIS RARE Barometer). "Time to diagnosis and determinants of diagnostic delays of people living with a rare disease." European Journal of Human Genetics 2024;32(9):1116-1126. Average time to diagnosis 4.7 years; 22 percent of patients (about 1 in 5) consult 8 or more health professionals.
10. **Shire Rare Disease Impact Report, 2013.** Time to diagnosis 7.6 years (United States) and 5.6 years (United Kingdom); a patient typically sees 8 physicians and receives 2 to 3 misdiagnoses.
11. **Government of India, Ministry of Health and Family Welfare.** National Policy for Rare Diseases, 2021. Three-group framework; 63 notified diseases; treatment support up to Rs 50 lakh per patient at Centres of Excellence.
12. **Government of India, Lok Sabha.** Written reply of the Minister of State for Health, 6 February 2026. India operates 15 Centres of Excellence for rare diseases (up from 8 at the policy's 2021 inception), including AIIMS Patna (designated by Office Memorandum dated 7 August 2025).
13. **Government of India, Lok Sabha.** Written reply of the Minister of State for Health, 9 August 2024 (Press Information Bureau, release PRID 2043516). 1,118 patients had benefited under the policy since March 2021.
14. **Indian Council of Medical Research.** National Registry for Rare and Other Inherited Disorders (NRROID). Enrollment of 15,369 patients across 23 centres and 231 diseases as of February 2025 (2025 registry report).
15. **The Hindu, February 2026** (based on a Right to Information reply). Of about Rs 299 crore allocated for rare-disease treatment in 2025-26, only about Rs 30.79 crore was utilised, with roughly Rs 271 crore set to lapse on 31 March 2026.
16. **High Court of Delhi**, order of 4 October 2024, Master Arnesh Shaw v. Union of India (directing a Rs 974 crore National Fund for Rare Diseases); stayed by the Supreme Court of India, 9 December 2024, with the matter pending.
17. **Government of India, Union Budget 2026-27** (Press Information Bureau). Ministry of Health and Family Welfare allocation Rs 1,06,530 crore; Indian Council of Medical Research Rs 4,000 crore; seven additional rare diseases granted customs-duty exemption on imported therapies.
18. **Government of India.** National Sickle Cell Anaemia Elimination Mission, 2023. Bihar among high-burden states; documented tribal sickle-cell prevalence.

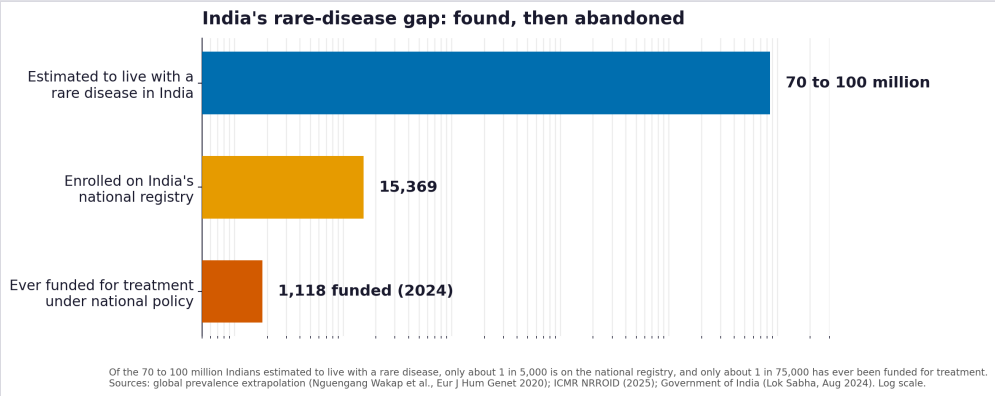
19. **Anjana RM, et al.** (ICMR-INDIAB). "Metabolic non-communicable disease health report of India." Lancet Diabetes and Endocrinology 2023. National multi-stage, community-based epidemiology across 31 states and union territories; the design precedent for a national rare-disease prevalence study.
20. **Thalassaemia International Federation.** Guidelines for the Management of Transfusion-dependent Thalassaemia, 4th edition, 2021. Survival with treatment; gene therapy (betibeglogene autotemcel).
21. **World Federation of Hemophilia.** Guidelines for the Management of Hemophilia, 3rd edition, 2020. Factor replacement, emicizumab, and gene therapy.
22. **Birnkrant DJ, et al.** "Diagnosis and management of Duchenne muscular dystrophy." Lancet Neurology 2018 (three-part care considerations). Corticosteroids, respiratory and cardiac care; exon-skipping and gene therapy.
23. **Butler MG, Manzardo AM, Heinemann J, Loker C, Loker J.** "Causes of death in Prader-Willi syndrome." Genetics in Medicine 2017;19(6):635-642. Mean age at death 29.5 years; 70 percent of deaths in adulthood.
24. **Landfeldt E, et al.; Broomfield J, et al.** (UK CPRD, Brain and Behavior 2023). Duchenne muscular dystrophy life expectancy historically late teens to early twenties, extending toward the late twenties to forties with modern care.
25. **National Health Mission, India (2016); Colah RB, et al., Pediatric Hematology Oncology Journal 2017.** Guidelines for Prevention and Control of Hemoglobinopathies in India; carrier frequency 3 to 4 percent; roughly 10,000 to 15,000 thalassemia-major-affected births per year (a 2023 meta-analysis by Dharmarajan and Kar implies about 8,700 affected births per year).
26. **Orphanet / Orphadata; NIH GARD; NHLBI; American Society of Hematology (2020).** Per-disease prevalence figures and disease-specific clinical guidelines used in the disease reference table.

ONE-PAGE BRIEFING

Rare diseases in India, in one page

1 in 10 people lives with a rare disease

In the United States, which counts, 1 in 10 people has a rare disease (NIH, NORD). Worldwide, over 300 million people live with more than 7,000 rare diseases (WHO, 2025). India has never counted.



15

Centres of Excellence for rare diseases in all of India, including AIIMS Patna (Government of India, 2026).

1,118

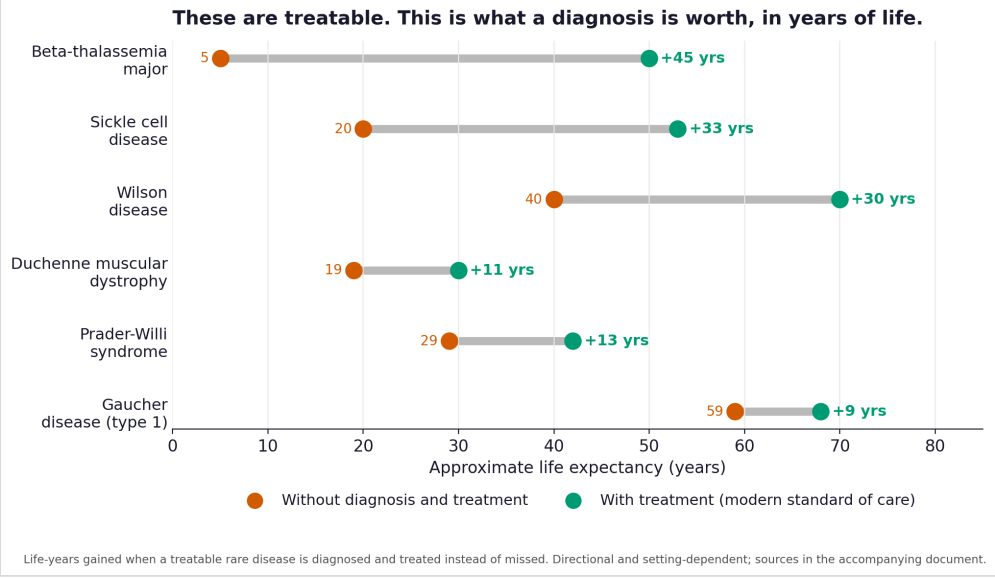
Patients ever funded for treatment under the national policy since 2021, out of tens of millions estimated (Government of India, 2024).

Rs 271 cr

Rare-disease treatment funds set to lapse unspent in 2025-26, because patients are not being found.

70-100 M

Indians estimated to live with a rare disease, a number never measured in India.



Life-years lost is preventable. Considering thalassemia alone, and using conservative assumptions (roughly one third of 10,000 to 15,000 affected births each year going undiagnosed or without adequate treatment, each losing about 40 years of life), India loses on the order of 130,000 to 200,000 years of life every year from a single treatable disease. This is an illustration, not a measurement, because India has never counted. That is the point.

Rare diseases by the numbers: India and the world

The world

Fact	Figure	Source
People living with a rare disease, worldwide	Over 300 million	WHO 2025; UN 2021[1,2]
Number of distinct rare diseases	More than 7,000	WHO 2025[1]
Share of the population affected (point prevalence)	3.5 to 5.9 percent	Nguengang Wakap 2020[3]
Rare diseases that begin in childhood	About 70 percent	WHO 2025[1,3]
Rare diseases with no approved disease-modifying treatment	About 95 percent	NIH/NCATS; Lancet Glob Health 2024[4,8]
United States: share of people with a rare disease	1 in 10 (25 to 30 million)	NIH/NCATS; NORD[4,5]
Europe: people with a rare disease	About 30 million (6 to 8 percent over a lifetime)	EURORDIS[6]
Average time from first symptom to diagnosis	About 4.7 years (up to 7.6 years)	EURORDIS 2024; Shire 2013[9,10]
Children with a rare disease who die before age 5 (community estimate)	About 30 percent	Cited by Lancet Glob Health 2024[8]

India

Fact	Figure	Source
Indians estimated to live with a rare disease (extrapolated)	70 to 100 million	Applying ref. 3 to India
A measured national prevalence of rare diseases	None exists	See ref. 3, 19
Diseases notified for support under national policy	63 (of more than 7,000)	NPRD 2021[11]
Government Centres of Excellence	15 (from 8 in 2021)	Lok Sabha, Feb 2026[12]
Patients ever funded for treatment	1,118 (since 2021)	Lok Sabha, Aug 2024[13]
Patients on the national registry	15,369	ICMR NRROID, Feb 2025[14]
Rare-disease treatment funds lapsing unspent, 2025-26	About Rs 271 crore	The Hindu (RTI), Feb 2026[15]
Thalassemia-major-affected births per year	About 10,000 to 15,000	NHM 2016; Colah 2017[25]
Proven national capacity for large community studies	ICMR-INDIAB, 31 states/UTs	Anjana 2023[19]

Bihar and the eastern states. AIIMS Patna is now Bihar's Centre of Excellence for rare diseases, with an antenatal diagnostic facility.[12] Bihar is a high-burden state under the National Sickle Cell Anaemia Elimination Mission.[18] A state expanding its medical colleges is well placed to lead, by ensuring central funds reach patients at AIIMS Patna rather than lapsing.

The most common rare and orphan diseases: what a diagnosis is worth

Ordered by their burden in India. "Age at diagnosis" compares Western countries with India, where diagnosis is consistently later. Where no national Indian figure exists, that is stated: it is itself evidence of the counting gap. Treatments that change the course of the disease are marked in green.

Disease	How common	Diagnosed at (West / India)	Life without treatment	Life with treatment	Treatment
Beta-thalassemia (a blood disorder)	India's largest burden; carriers 3 to 4 percent; 10,000 to 15,000 affected births a year	Infancy / often only after an affected birth	Death in early childhood	Majority live past 50 to 60	**YES: transfusion and chelation; bone-marrow transplant; gene therapy**
Sickle cell disease (a blood disorder)	Very high in tribal communities (carrier up to a third in some)	Birth (screening) / late, often symptomatic	High childhood death	Into adulthood (about 50s with full care)	**YES: hydroxyurea; transplant; gene therapy**
Hemophilia (a bleeding disorder)	About 140,000 Indians, second most in the world; only about 26,000 registered	Early / around age 5	Death in childhood or early adulthood from bleeding	Near-normal life	**YES: clotting-factor therapy; emicizumab; gene therapy**
Duchenne muscular dystrophy (muscle wasting)	About 1 in 5,000 boys	Age 4 to 5 / later	Death in the late teens or early twenties	Into the late twenties to forties	**YES: steroids; exon-skipping; gene therapy**
Spinal muscular atrophy (muscle weakness)	About 1 in 10,000	Newborn screening / symptomatic	Severe form: death by about age 2	Survival and milestones transformed	**YES: nusinersen; risdiplam; one-time gene therapy**
Cystic fibrosis (lungs and digestion)	Underdiagnosed in India	2 to 4 months / 2 to 10 years	Early death in childhood	Into the 40s and 50s	**YES: CFTR modulator therapy (access-limited)**
Wilson disease (copper build-up)	India a hotspot; about 1 in 18,700 (genome-based estimate)	Childhood to young adult / delayed about 2 years	Fatal from liver or brain damage	Near-normal life	**YES: chelation and zinc (highly treatable)**
Phenylketonuria (a metabolic disorder)	About 1 in 10,000 to 25,000 (limited Indian screening)	At birth (screening) / often missed	Severe, permanent intellectual disability	Normal intelligence and lifespan	**YES: special diet from birth (fully preventable)**
Congenital adrenal hyperplasia (a hormone disorder)	About 1 in 14,000 to 18,000	Newborn screening / often at crisis	Fatal salt-losing crisis in newborns	Normal life	**YES: hydrocortisone (life-saving)**

Disease	How common	Diagnosed at (West / India)	Life without treatment	Life with treatment	Treatment
Pompe disease (a storage disorder)	About 1 in 18,700	Under 6 months (severe) / delayed	Severe form: death around age 1	Improved survival with early therapy	**YES: enzyme replacement therapy**
Gaucher disease (a storage disorder)	The most common storage disorder in India	Childhood to adult / delayed	Progressive organ and bone disease	Near-normal (type 1)	**YES: enzyme replacement and substrate therapy**
Fabry disease (a storage disorder)	About 1 in 15,000	Childhood (classic) to adult / delayed	Life shortened 10 to 20 years (kidney, heart, stroke)	Progression slowed	**YES: enzyme replacement; oral chaperone**
Mucopolysaccharidosis I and II (storage disorders)	About 1 in 100,000; second-largest storage group in India	Infancy to early childhood / delayed	Death in the first or second decade	Slowed with therapy (brain effects limited)	**YES: enzyme replacement; transplant**
Prader-Willi syndrome (a genetic syndrome)	About 1 in 15,000 to 30,000	Infancy / delayed	Early death from obesity, around the late 20s	Into the 40s and beyond	**PARTIAL: growth hormone and strict food control**
Tuberous sclerosis (tumor-predisposition)	About 1 in 6,000 to 10,000	Infancy or childhood / delayed	Seizures, organ tumors, disability	Much improved with surveillance	**YES: mTOR inhibitor therapy**
Marfan syndrome (connective tissue)	About 1 in 5,000	Any age (delay 5 to 7 years) / delayed	Premature death from aortic rupture	Near-normal with monitoring	**YES: medication and preventive heart surgery**
Osteogenesis imperfecta (brittle bones)	About 1 in 10,000 to 20,000	Birth to childhood / delayed	Severe form fatal at birth; milder forms disabling	Fewer fractures, better mobility	**PARTIAL: bone-strengthening medicines and surgery**
Neurofibromatosis type 1 (nerve tumors)	About 1 in 3,000	Childhood / delayed	Variable; some tumors turn dangerous	Surveillance; targeted therapy for some	**PARTIAL: selumetinib for specific tumors**
Huntington disease (a brain disorder)	Less common in India than the West	Adulthood (ages 30 to 50)	Death about 15 to 20 years after onset	Symptoms eased; course not yet altered	**NO disease-modifying therapy yet**
Fragile X syndrome (intellectual disability)	About 1 in 4,000 boys	Early childhood / delayed	Lifelong intellectual disability (not life-shortening)	Function improved with therapy	**NO disease-modifying therapy yet**

The pattern is unmistakable. Most of the diseases that affect the most Indians are treatable, and several are treatable cheaply. What is missing is not the medicine. What is missing is the diagnosis. Every "delayed" in the India column is a child found too late.

THE PETITIONERS

We, the undersigned, ask the Government to count and care for India's rare-disease patients

This petition is offered for signature by the organizations, physicians, scientists, and families of India's rare-disease community, and by international partners who stand with them. The organizations below are invited as founding cosignatories; their listing here indicates the intended roster, and each signs in its own name.

Indian national umbrella organizations: Organization for Rare Diseases India (ORDI); Indian Organization for Rare Diseases (IORD); Foundation for Research on Rare Diseases and Disorders (FRRDD); Rare Diseases India Foundation; Centre for Health Ecologies and Technology (CHET).

Indian patient organizations: Thalassemics India; Federation of Indian Thalassemics; Thalassemia and Sickle Cell Society (Hyderabad); Sickle Cell Society of India; Hemophilia Federation (India); Cure SMA Foundation of India; Dystrophy Annihilation Research Trust (DART); Indian Association of Muscular Dystrophy; Lysosomal Storage Disorders Support Society (LSDSS); Metabolic Errors and Rare Diseases Organisation of India (MERD India); Indian Prader-Willi Syndrome Association; Down Syndrome Federation of India; Indian Rett Syndrome Foundation; Fragile X Society India; Niemann-Pick India; World Without GNE Myopathy.

International organizations invited to endorse: Rare Diseases International (which led the 2021 UN Resolution); EURORDIS-Rare Diseases Europe; National Organization for Rare Disorders (NORD, USA); Global Genes; Thalassaemia International Federation; World Federation of Hemophilia; World Duchenne Organization; International Prader-Willi Syndrome Organisation; Asia Pacific Alliance of Rare Disease Organisations (APARDO); Beacon for Rare Diseases.

Physicians, medical geneticists, and scientists from India's Centres of Excellence and medical-genetics departments, and the members of the ICMR Rare Diseases National Consortium, are invited to co-sign in their individual capacities.

Signatures

Name:

Title and organization:

City and State:

Signature and date:

Name:

Title and organization:

City and State:

Signature and date:

Name:

Title and organization:

City and State:

Signature and date:

Name:

Title and organization:

City and State:

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Additional signature pages may be appended. Each cosignatory consents to the public listing of their name and organization in support of this petition.