A Typology and Design Framework for National Rare Disease Policies and Plans

A diagnostic and design-support tool for policymakers and their partners

Global Rare Disease Policy Network | Rare Toolbox

Working draft — July 2026

Executive Summary

Rare diseases (RDs), though individually uncommon, collectively affect an estimated 6 to 8 percent of people worldwide. Most are genetic, many begin in childhood, and many are severe or life-shortening. Over the past two decades a growing number of governments have responded with national RD laws, strategies, and programs. The result is a fast-expanding but uneven and hard-to-navigate policy landscape, particularly across the Network's priority regions of Asia Pacific and Latin America, where economies sit at very different levels of health-system maturity.

A policymaker asked to design, revise, or defend a national RD plan faces a practical difficulty: there is no shared, neutral map of the design choices a plan involves, the trade-offs each choice carries, or the sequence in which a plan can realistically be built. Existing comparative work does not fill this gap. It is largely descriptive, region-specific, and point-in-time, and it classifies what economies have done rather than helping decide what to do next. A 2025 scoping review of RD policy-evaluation frameworks identified only five, and concluded that structured, decision-oriented instruments remain scarce.

This tool responds to that gap. It reframes the typology of national RD plans from a descriptive classification into a design and diagnostic instrument. It has four parts:

1. **Foundational archetypes** (Part A) locate the overall shape of a plan on two axes: how it is legally anchored, and how it is integrated into the wider health system.
2. **Design components** (Part B) treat the nine building blocks every plan must decide on. Each is presented as a decision with a spectrum of options, the trade-offs involved, the conditions favoring each option, real examples, and a marker distinguishing a minimum-viable from an advanced approach.
3. **Maturity and sequencing** (Part C) set out a staged pathway so that economies with limited resources can identify a credible next step rather than an unreachable ideal.
4. **Self-diagnostic** (Part D) converts the framework into a worktool: a short set of decision questions and a gap-analysis template that turn a reading of the paper into a prioritized design agenda.

What this tool is not. It is not a ranking of governments, a compliance checklist, or a single recommended model. Economies differ in fiscal space, legal tradition, health-system design, and political context, and strong plans exist in many different forms. The tool is a shared vocabulary and a menu of informed choices, aligned with the voluntary, consensus-based spirit of the APEC Action Plan on Rare Diseases, of which the Network is steward.

How it fits the Toolbox. This paper is the design-side companion to the Network's other instruments. Its components map directly to the ten pillars of the APEC Action Plan, and it is designed to be used alongside the Network's 30-indicator scoring rubric, which assesses implementation. The two treatments of access to therapies in the Toolbox, on regulatory review and on health technology assessment (HTA) and reimbursement, are treated here as components rather than re-derived.

1. Introduction: purpose and how to use this tool

1.1 The problem this tool addresses

The case for national RD policy is now well established. Coordinated national action shortens the diagnostic odyssey, improves equity of access, and generates wider co-benefits for health systems, from stronger newborn screening and genomic infrastructure to advances that benefit more common conditions. What is missing is not the argument for action but a practical means of translating that argument into a well-designed plan.

Two audiences feel this gap most directly. Government policymakers must make concrete choices about governance, financing, data, diagnosis, access, and delivery, often without a structured way to weigh the options or to see how comparable economies have handled the same decisions. Their partners in the RD community, including clinician key opinion leaders (KOLs) and patient-organization leaders, must persuade government and co-design the plan, but frequently lack a common language in which to frame proposals. When the two sides argue from different mental models, negotiations stall and plans emerge incomplete.

1.2 Audience and use cases

- **Government policymakers** drafting or revising a national RD policy, plan, or law, or preparing a business case for finance and cabinet colleagues.
- **KOL and patient-advocacy partners** seeking to influence government and to co-design specific components of a plan.
- **Regional bodies and reviewers** benchmarking progress, planning capacity-building, or coordinating peer learning across economies.

1.3 What this tool is and is not

The tool is a shared design map and a structured diagnostic. It is deliberately not a scorecard, not a ranking, and not an endorsement of one model over others. Placing an economy in an archetype or noting a gap describes structure, not merit; a minimum-viable plan pursued honestly may serve patients better than an ambitious plan that is unfunded or unimplemented. Readers should treat every option in Part B as legitimate under the right conditions, and should read Part C as a menu of feasible next steps rather than a hierarchy of worth.

1.4 Scope, method, and evidence base

The tool draws primarily on national plans, laws, and program documents from Asia Pacific and Latin America, consistent with the Network's mandate, with reference to high-income systems where they illustrate a design choice clearly. The illustrative material was assembled from official government and regulator sources, multilateral publications, peer-reviewed literature, and Network stakeholder input, and cross-checked where possible.

Evidence limitations, stated plainly. Coverage is uneven. Many plans exist only in local languages or in gray literature, and the gap between a plan on paper and its implementation is frequently undocumented. Point-in-time figures such as budget sizes, numbers of listed diseases or reimbursed products, and screening panel counts change with annual cycles and are treated cautiously here; where a specific number would date quickly, the paper describes the structural feature rather than the figure. The examples are meant to illustrate design choices, not to certify the current status of any economy.

2. Framing: national RD policy as a set of design choices

2.1 A working definition

A national RD policy or plan is defined here inclusively: any coordinated national framework, whether a dedicated law, a standalone strategy, or a distinct component of a broader health framework, that sets out to improve the diagnosis, treatment, and social support of people living with rare diseases. The prevalence threshold used to define a rare disease varies across jurisdictions, and a globally applicable operational description of one in 2,000 people

within a WHO region has been proposed but is not universally adopted. The tool does not depend on a single definition; it applies wherever an economy is trying to organize a coherent response.

2.2 The core problem every plan is trying to solve

Beneath the variation, every national plan addresses the same structural problem. Patients are few per disease and dispersed across the population, so the default health system underdiagnoses them, struggles to concentrate expertise, and finds their care difficult to finance and to measure. A plan is the mechanism by which an economy overcomes that structural disadvantage through deliberate coordination. Framing the task this way clarifies why the design components in Part B are common to all plans even when the archetype in Part A differs.

2.3 Alignment with the APEC Action Plan on Rare Diseases

The APEC Action Plan on Rare Diseases, endorsed by APEC Health Ministers in 2018 and stewarded by the Network, organizes RD policy around a Vision for 2025, three objectives, and ten pillars, with associated targets and indicators. The ten pillars are: (1) define rare diseases and orphan products; (2) raise public and political awareness; (3) promote innovative research and development; (4) build human-resource capacity; (5) facilitate early, accurate, and systematic diagnosis; (6) coordinate patient-centered care; (7) deliver new and accessible treatments; (8) support the financial and social needs of patients and families; (9) manage the pooling and use of patient data securely; and (10) prioritize a comprehensive domestic RD policy integrating pillars one through nine.

This tool is built to be consistent with, and to operationalize, that structure. The design components in Part B map onto the pillars (see Annex B), so that an economy using the APEC framework can move directly from the pillars to concrete design choices, and can pair this design-side tool with the Network's 30-indicator scoring rubric on the assessment side.

2.4 Why a new tool is needed

Useful comparative work already exists, but none of it does the job this tool sets out to do. Normative anchors such as the 2021 UN resolution on persons living with a rare disease and the WHO and ICD-11 definitional work set direction but do not classify or guide the design of plans. Trackers and resource maps from Orphanet, Rare Diseases International, and EURORDIS are descriptive inventories, often region-specific and snapshot-based. Academic reviews, including a 2017 comparison of eleven national policies against patient-need dimensions, a 2018 compilation across twenty-three countries, a 2022 scoping review of RD health policies, and a 2024 comparison of funding approaches in five high-income systems, are point-in-time, sample-limited, and not maintained, and most describe status and gaps rather than offering transferable decision guidance. A 2025 scoping review of RD policy-evaluation frameworks found only five and concluded that structured instruments are lacking. The niche this tool occupies is therefore a decision-oriented, globally applicable, and maintainable design framework, explicitly mapped to the APEC pillars and intended to be updated as a living resource rather than fixed in a single publication.

3. Part A — Foundational archetypes (orientation typology)

Before examining individual components, it helps to locate the overall shape of a plan. Rather than the eight overlapping dimensions used in earlier drafts, Part A uses two axes that capture the most consequential structural choices. The archetype is an orientation aid, not a verdict: strong and weak plans exist within every cell, and many economies are in transition between cells.

3.1 Axis 1 — Legal anchoring

How is the plan authorized? A legislation-based plan is codified in primary law, which confers durability, enforceable entitlements, and protection across political cycles, at the cost of being slower to enact and harder to adapt. A policy-guided plan rests on strategy documents or administrative decisions, which are faster and more flexible but depend on sustained political will. A hybrid approach combines a legal backbone with adaptable implementing programs, and is often the most robust arrangement in practice.

3.2 Axis 2 — Integration

How does the plan relate to the wider health system? A standalone plan addresses rare diseases as a discrete agenda, which maximizes visibility and dedicated attention but risks operating in parallel to mainstream services. An embedded plan places rare diseases within a broader framework such as universal health coverage, which promotes sustainability and system leverage but can dilute dedicated focus and funding. A networked variant coordinates rare-disease action across several existing programs without a single home.

3.3 The archetype map

The exhibit below places illustrative economies on the two axes. Placement reflects the structural anchoring of the plan and implies nothing about the quality of implementation. Several economies could sit across a boundary; they are shown where their principal anchoring lies.

Integration \ Legal anchoring	Legislation-based	Policy-guided	Hybrid
Standalone	Chinese Taipei; Republic of Korea; Japan; Argentina; Peru	Australia	Chile (framework law over pre-existing financing)
Embedded / networked	Philippines; Colombia	Brazil; Malaysia; Thailand; Singapore	People's Republic of China (lists and notices within general regulation)

Two patterns are worth noting for the discussion that follows. First, several economies with dedicated laws still deliver most access through the general health system, so a strong legal anchor does not remove the component choices in Part B; it shapes but does not settle them. Second, the policy-guided and embedded cell is where much recent movement is concentrated, as economies add explicit RD provision to existing frameworks. Archetype, in short, is a starting point for the real work of component design.

4. Part B — Design components (the working framework)

This is the core of the tool. Each of the nine components below is a decision that every plan must make, whatever its archetype. Each is treated with the same structure: the decision and why it matters, the spectrum of options and their trade-offs, illustrative examples, the corresponding APEC pillar, and a marker distinguishing a minimum-viable from an advanced approach. The components interact, and a coherent plan aligns its choices across them; the ordering here is analytical, not a sequence of priority. Part C addresses sequencing.

4.1 Governance and coordination

The decision. Who leads the plan, and how is action coordinated across the institutions and stakeholders that must contribute? Rare diseases cut across regulators, payers, clinical services, research, and social sectors, so an explicit coordination mechanism is usually the difference between a plan that moves and one that fragments.

Options and trade-offs. Options range from a single named lead agency with a light coordinating function, through a standing cross-ministry mechanism, to a statutory multi-stakeholder body with formal patient and clinician seats. Centralized coordination brings uniformity and accountability but can be distant from local needs; multi-stakeholder governance improves buy-in and expertise but slows consensus and demands staff time to run well.

Illustrative examples. Japan coordinates through the health ministry with a statutory advisory council and long-standing patient representation. Malaysia established a National Rare Disease Committee in 2019 with technical sub-committees and confirmed patient-organization seats. Colombia's Mesa Nacional de Enfermedades Huérfanas institutionalizes patient and family seats alongside scientific and payer representation, though its role is advisory.

Minimum viable: a named lead official or unit with a basic cross-ministry coordination routine and a defined channel for patient input.

Advanced: a mandated multi-stakeholder governance body with defined membership, patient and clinician seats, conflict-of-interest rules, and a published work program.

APEC pillar: *Pillars 6 and 10*

4.2 Scope and multisectoral reach

The decision. Does the plan address only health-sector functions, or does it reach across government into education, labor, social protection, and disability services? Much of the burden of rare disease falls outside clinical care, so scope determines whether a plan meets patients' actual needs.

Options and trade-offs. A health-sector-focused plan is simpler to govern and finance but leaves major needs unmet. A whole-of-government plan addresses education, employment, income support, and disability, at the cost of harder coordination and shared accountability. A practical middle path anchors the plan in health while creating explicit bridges to social entitlements.

Illustrative examples. The Philippines' Rare Diseases Act assigns tasks across seven agencies and, distinctively, recognizes persons with a rare disease as persons with disability, opening statutory social-support channels. Malaysia routes support through its disability (OKU) framework. Many plans remain predominantly health-sector in practice even where broader intent is stated.

Minimum viable: a health-sector plan with at least one defined link to existing disability or social-support entitlements.

Advanced: a whole-of-government plan with named responsibilities across education, labor, and social protection and a mechanism to hold them jointly accountable.

APEC pillar: *Pillars 6, 8, and 10*

4.3 Definitions, case ascertainment, and data

The decision. How does the economy define a rare disease, identify cases, and build the data infrastructure on which every other function depends? Without an agreed definition and a usable registry, planning, financing, and evaluation all rest on guesswork.

Options and trade-offs. Choices span the definition itself (numerical threshold, official disease list, or alignment with ICD-11), and the data architecture (from a minimum dataset on priority conditions to interoperable registries feeding regulatory, HTA, and reassessment decisions). Registries are consistently the weakest layer across both priority regions; the recurring failure is a registry that is a descriptive repository with no decision use.

Illustrative examples. Chile's 2025 framework law mandates an official disease list and a national registry, with implementing standards approved in 2026. Colombia operates one of the region's more established registries, integrated with routine surveillance systems. Several economies mandate registries in law but have not made them operational, a gap the tool flags rather than papers over.

Minimum viable: an agreed working definition and a minimum dataset for a small number of priority conditions.

Advanced: interoperable, governed registries built on common standards that feed regulatory, HTA, and reassessment decisions and support cross-border pooling for ultra-rare conditions.

APEC pillar: *Pillars 1 and 9*

4.4 Diagnosis

The decision. How does the plan shorten the diagnostic odyssey through newborn and genetic screening and organized diagnostic networks? Diagnosis is the entry point to all subsequent care, and it is the layer most consistently developed across the priority regions.

Options and trade-offs. Options include the breadth of the newborn screening panel, whether expanded and genomic screening are publicly funded and universal or pilot and regional, and whether diagnostic expertise is networked through centers or left to chance. Broader panels and genomic diagnosis find more patients earlier but raise cost, workforce, and counseling demands, and equity suffers where access is concentrated in capital cities.

Illustrative examples. Newborn screening is near-universal in several APAC economies and expanding elsewhere; Chile expanded its panel substantially on the way to universal coverage, and Korea moved newborn metabolic and hearing screening to near-universal public coverage. Undiagnosed-disease and genomic programs have been established in Japan, Korea, and elsewhere.

Minimum viable: mandatory core newborn screening and a defined referral route for suspected cases to specialist expertise.

Advanced: publicly funded expanded and genomic screening with organized diagnostic networks and equitable geographic access.

APEC pillar: *Pillar 5*

4.5 Access to therapies (regulation, HTA, reimbursement, financing)

The decision. How do patients actually obtain therapies once they exist? This spans regulatory review and orphan designation, health technology assessment, reimbursement listing, and the financing that pays for treatment. It is the most contested and resource-intensive component.

Options and trade-offs. The Network's Toolbox treats this domain in depth in two dedicated papers, on optimizing regulatory review and on recognizing and rewarding value through HTA and reimbursement. Rather than re-derive them, this tool treats access as a component and points to those papers for the detailed options: adaptive and reliance-based regulatory pathways, RD-adapted HTA with severity and unmet-need modifiers, and financing models discussed in the next component. The central trade-off is between speed of access and management of clinical and budgetary uncertainty.

Illustrative examples. Regional collaboration such as Project Orbis, the ACCESS Consortium, and ASEAN Joint Assessment shows how work-sharing and reliance shorten timelines for smaller markets. Latin American systems generally handle orphan products through general medicines regulation, HTA, and reimbursement rather than a dedicated designation regime.

Minimum viable: a defined route by which orphan products can be authorized and funded, using reliance on trusted reference authorities where local capacity is limited.

Advanced: dedicated, proportionate regulatory and HTA pathways with life-cycle evidence and managed-entry arrangements, coordinated regionally.

APEC pillar: *Pillar 7*

4.6 Financing and sustainability

The decision. How is rare-disease care paid for, and is the financing durable? A plan without a credible and sustained financing model is the most common and damaging failure mode, whatever its other strengths.

Option	Fits when...	Key trade-off
Dedicated RD fund	Political salience is high and ring-fencing is feasible	Protected and focused, but often capped and vulnerable at review
Integrated financing (UHC / insurance)	Strong pooled financing already exists	Equitable and durable, but high-cost therapies need explicit provisions
Public-private / pooled	Fiscal space is tight and partners are willing	Adds resources, but needs governance to protect the public interest

Options and trade-offs. The main options are shown below. Most economies combine elements, and the decisive question is less the label than whether financing is protected, sufficient, and stable across budget cycles.

Illustrative examples. Chile's Ley Ricarte Soto is a dedicated high-cost fund covering decreed therapies for all insurers, though its expansion has stalled and it faced an acute financing strain in 2025 to 2026, illustrating the vulnerability of dedicated funds at review. Singapore's Rare Disease Fund uses three-to-one government matching of donations, a public-private model. Japan, Korea, and Chinese Taipei relieve cost through statutory co-payment mechanisms layered on universal insurance.

Minimum viable: a defined budget line or partner-supported financing for a small set of priority conditions.

Advanced: sustainable pooled financing with explicit provisions for high-cost therapies and a reassessment cycle that adjusts over time.

APEC pillar: *Pillar 8*

4.7 Patient and caregiver engagement

The decision. How, and how early, are patients and caregivers involved in designing, delivering, and evaluating the plan? Engagement determines whether a plan reflects lived priorities or only institutional assumptions.

Options and trade-offs. Options run from consultation-only involvement at the end of a process, through structured participation across the policy cycle, to formal roles on governance and appraisal bodies. Genuine co-design surfaces needs and builds trust but requires governments to share influence and to invest time; consultation-only is faster but risks missing what matters and eroding confidence.

Illustrative examples. Rare Voices Australia functions as a recognized national peak body that co-developed the national plan. FADEPOF in Argentina and FECOER in Colombia hold formal seats on advisory bodies. Across both regions, patient organizations have repeatedly driven the adoption of specific benefits, though formal governance seats remain uneven.

Minimum viable: a defined, regular channel for patient and caregiver input into the plan.

Advanced: formal patient and caregiver roles across governance, appraisal, and evaluation, with support for organizations to build data and advocacy capacity.

APEC pillar: *Pillars 2 and 8*

4.8 Service delivery and implementation

The decision. How is care organized and delivered, and how is the plan rolled out? This covers centers of excellence and referral networks, workforce capacity, and whether implementation is phased, comprehensive, or piloted before scale-up.

Options and trade-offs. Delivery choices include concentrating expertise in designated centers versus distributing it, and building workforce capacity deliberately versus relying on existing specialists. Rollout can be phased (lower risk, slower benefit), comprehensive (fast but hard to sustain), or pilot-to-scale (prudent but dependent on genuine evaluation and the will to expand). A common failure is a pilot that is never scaled.

Illustrative examples. Clinical networks and designated centers coordinate care in several economies, including national collaboration networks and centers of expertise. Workforce shortages, especially of clinical geneticists, and concentration of specialists in capital cities are recurring constraints across both regions.

Minimum viable: at least one designated center or referral pathway and a realistic phased rollout plan.

Advanced: a networked system of centers of excellence with deliberate workforce development and a tested pilot-to-scale mechanism.

APEC pillar: *Pillars 4 and 6*

4.9 Monitoring, evaluation, and learning

The decision. How will the plan know whether it is working, and how will it improve? Monitoring choices determine whether a plan can be adjusted, defended at budget time, and learned from.

Options and trade-offs. Options range from embedding rare diseases in general health monitoring with no RD-specific indicators, to a dedicated M&E framework with targets, indicators, and a scheduled reassessment cycle. Dedicated M&E enables accountability and course-correction but requires data infrastructure and analytic capacity; embedded monitoring is lighter but often renders rare diseases invisible in the numbers.

Illustrative examples. The APEC Action Plan's own indicator structure, and the Network's 30-indicator scoring rubric, provide a ready set of measures an economy can adopt or adapt rather than build from scratch.

Minimum viable: a small set of decision-relevant indicators and a defined review point.

Advanced: a dedicated M&E framework aligned to the APEC indicators, with scheduled reassessment feeding back into financing and design.

APEC pillar: *Pillars 9 and 10*

5. Part C — Maturity and sequencing

A plan is built over time, not adopted whole. The most common mistake in RD policy advice is to hold up the most elaborate systems as the standard for economies that lack the fiscal space, workforce, or data to reach them, which produces either paralysis or unfunded mandates. This part sets out a staged pathway so that any economy can identify a credible next step. The stages describe what is in place across the Part B components, not the worth of the economy pursuing them.

5.1 A three-stage pathway

The stages are minimum viable, developing, and advanced. An economy will often sit at different stages on different components, which is expected; the pathway is a way to prioritize, not a single ladder. The table illustrates the progression on three representative components.

Component	Minimum viable	Developing	Advanced
Governance	Named lead and basic coordination	Cross-ministry mechanism with patient input	Statutory multi-stakeholder body with patient and clinician seats
Data	Minimum dataset on priority conditions	National registry operational for core conditions	Interoperable registries feeding HTA and reassessment
Financing	Line item or partner-supported pilot	Defined reimbursement route within UHC or a fund	Sustainable pooled financing with high-cost provisions

5.2 Where to start when starting near zero

For an economy beginning with little in place, the evidence and the Network's experience point to a small number of foundational moves that unlock the rest: adopt a working definition; name a lead and a coordination routine; stand up a minimum dataset on a handful of priority conditions; secure a first, even modest, financing route, using reliance on trusted regulators to avoid duplicating capacity the economy does not yet have; and build on the strongest existing layer, which is usually newborn screening. This sequence mirrors the minimum-viable logic the Network applies in its regulatory and HTA papers, and it is more credible than attempting to replicate an advanced system at once.

5.3 How plans stall (common failure modes)

- **Unfunded mandates.** A law or strategy that creates obligations without a financing route, so the plan exists on paper only.
- **Laws without implementing rules.** Primary legislation that is never operationalized through regulations, as seen in the multi-year gap between enactment and financing in several economies.
- **Registries with no decision use.** Data collection that never informs regulatory, reimbursement, or planning decisions and so is not sustained.
- **Pilots that never scale.** Successful demonstrations that lack the political will or budget to expand nationally.

Anticipating these failure modes at the design stage, and building in the financing route, implementing rules, decision use, and scale-up trigger from the start, is one of the highest-value uses of this tool.

6. Part D — Self-diagnostic and design agenda

The framework becomes a worktool when an economy applies it to itself. The diagnostic has three steps.

5. **Answer the decision questions.** For each Part B component, the team answers a short set of questions about current arrangements. Examples: Is there a named lead and a functioning coordination mechanism? Is there an agreed definition and an operational registry with decision use? Is there a defined, funded route to therapy access? Are patients involved beyond consultation? Is there a reassessment cycle?

6. **Locate each component on the maturity pathway.** Using Part C, the team marks each component as minimum viable, developing, or advanced, producing an honest internal picture of strengths and gaps.
7. **Convert gaps into a prioritized design agenda.** The team selects the next feasible step on the highest-priority gaps, guided by the failure modes in 5.3 and by fiscal and political constraints, and sequences them.

Pairing with the scoring rubric. This diagnostic supports the design of a plan and is intended for internal use by an economy and its partners. It is distinct from, and complementary to, the Network's 30-indicator APEC scoring rubric, which assesses implementation against the Action Plan. Used together, the tool helps decide what to build and the rubric helps measure what has been built. Neither is intended to rank economies against one another.

7. Cross-cutting design principles

The tool deliberately does not propose a single harmonized model. Economies differ too much, and the Network's position elsewhere in the Toolbox is that convergence should be around principles with context-specific adaptation, not around one universal template. The following principles are contingent: they indicate what to favor under stated conditions.

- **Anchor durable commitments in law where enforceability matters; implement through adaptable programs where flexibility matters.** A hybrid arrangement often captures both.
- **Design for sustainability from the outset.** Financing and staffing are not implementation details; an unfunded plan is the most common avoidable failure.
- **Build patient and caregiver engagement into design, not only consultation.** The cost is time and shared influence; the return is relevance and trust.
- **Make monitoring decision-relevant and scheduled.** Indicators that no one acts on are a cost without a benefit.
- **Sequence honestly.** A well-implemented minimum-viable plan is worth more than an ambitious plan that stalls.

8. Applying the framework: worked vignettes

The following vignettes are composite and illustrative. They show how the tool is used across maturity levels; they are not assessments of specific economies.

8.1 An advanced economy revising a mature plan

An economy with a dedicated law, established registries, and universal financing runs the diagnostic and finds its gap is not coverage but life-cycle learning: therapies approved under uncertainty are rarely reassessed, and financing decisions are not revisited as evidence matures. Its design agenda is narrow and specific: add scheduled reassessment (component 9) tied to managed-entry arrangements (component 5), and strengthen the decision use of existing registries (component 3). The archetype does not change; the refinement is at the component level.

8.2 A mid-maturity economy consolidating fragmented efforts

An economy has newborn screening, some reimbursed orphan products, and active patient organizations, but no coherent plan and no single coordinating home. The diagnostic shows strength on diagnosis and engagement and weakness on governance and data. Its agenda is to establish a lead and a cross-ministry mechanism (component 1), make a registry operational with defined decision use (component 3), and consolidate scattered financing into a defined route (component 6), moving several components from developing toward advanced while leaving the archetype embedded.

8.3 An economy starting near zero

An economy with little formal RD provision uses Part C's starting sequence. Rather than draft an ambitious law it cannot implement, it adopts a working definition, names a lead, builds a minimum dataset on a few high-burden conditions, secures a first financing route using regulatory reliance, and builds on its existing newborn screening.

Each move is minimum viable, achievable within current capacity, and designed to avoid the failure modes in 5.3. The plan is modest, funded, and real, and it establishes the foundation for later stages.

9. Conclusion

The value of a typology of national RD plans lies not in classifying what economies have done but in helping them decide what to do next. This tool reframes the typology accordingly: two orientation axes to locate a plan, nine components that turn policy into concrete design choices with honest trade-offs, a staged pathway that meets economies where they are, and a self-diagnostic that converts all of it into a prioritized agenda. It is deliberately aligned with the APEC Action Plan's ten pillars and paired with the Network's scoring rubric, and it is deliberately not a ranking or a single prescribed model.

Plans change, and a static document dates quickly. The Network intends this paper as the first version of a maintained resource, and invites policymakers and their partners to use the diagnostic and to contribute their national experience so that the examples, archetypes, and staging improve over time. The goal is practical: to help every economy, at whatever stage, design a rare-disease policy that is coherent, feasible, and centered on the people it serves.

Annex A — Comparison matrix: design components across exemplar economies

Illustrative and structural only; reflects the principal anchoring of each economy's approach, not current implementation status or quality. Point-in-time figures are omitted by design. To be expanded in production into the full reference table readers will use most.

Economy	Legal anchor	Integration	Financing model	Notable feature
Australia	Policy-guided	Embedded (UHC)	Integrated (PBS + LSDP)	Recognized national peak body co-developed the plan
Japan	Legislation-based	Standalone + embedded	Statutory co-pay relief on universal insurance	Mature designated-disease system and diagnostics
Rep. of Korea	Legislation-based	Standalone	NHI with special co-pay and income support	Coherent listing-to-benefits pipeline
Chinese Taipei	Legislation-based	Standalone	NHI co-pay exemption + statutory subsidy	Among the earliest RD laws (2000)
Philippines	Legislation-based	Embedded (UHC)	PhilHealth package + disability benefits	RD patients recognized as persons with disability
China (PRC)	Hybrid (lists/notices)	Embedded	Reimbursement list + sub-national funds	Official disease lists; national clinical network
Singapore	Policy-guided	Embedded (3M)	Rare Disease Fund (3:1 PPP)	Public-private matched charity fund
Malaysia	Policy-guided	Embedded	Trust fund + RD-specific HTA pathway	2025 national policy mapped to APEC pillars
Chile	Hybrid (2025 law)	Standalone over embedded	Dedicated high-cost fund (Ricarte Soto)	New framework law; registry standards 2026
Colombia	Legislation-based	Embedded (SGSSS)	Integrated coverage (ADRES)	Established registry; institutionalized patient seats
Argentina	Legislation-based	Embedded (multi-payer)	Mandated coverage across three sectors	Statutory coverage guarantee; patient council seat

Annex B — Crosswalk: design components to APEC Action Plan pillars

Design component (Part B)	Primary APEC pillar(s)
1. Governance and coordination	6 (coordinate care); 10 (comprehensive policy)
2. Scope and multisectoral reach	6; 8 (financial and social needs); 10
3. Definitions, case ascertainment, and data	1 (define RDs and orphan products); 9 (patient data)
4. Diagnosis	5 (early, accurate diagnosis)
5. Access to therapies	7 (deliver accessible treatments); with 3 (R&D)
6. Financing and sustainability	8 (financial and social needs)
7. Patient and caregiver engagement	2 (public and political awareness); 8
8. Service delivery and implementation	4 (human-resource capacity); 6
9. Monitoring, evaluation, and learning	9 (data); 10 (comprehensive policy)

Pillar 2 (awareness) and Pillar 3 (research and development) are cross-cutting and are engaged by several components; they can be added as standalone components in production if the Network prefers full pillar-by-pillar coverage.

Annex C — Method and evidence limitations

Illustrative material was assembled from official government and regulator publications, multilateral and peer-reviewed sources, and Network stakeholder input, and cross-checked where feasible. Legal and structural facts (the existence, name, year, and anchoring of laws and plans) are high-confidence. Lower-confidence items, flagged for verification before publication, include the operational status of newly established registries; newborn-screening implementation rates against legal mandates; the current status of pending legislation; and any point-in-time figure for budgets, listed diseases, reimbursed products, or screening panels. Inline attributions in this draft are indicative; full citations will be finalized in production, following the reference style of the Network's regulatory and HTA papers. Nothing in this paper should be read as certifying the current implementation status of any named economy.

Selected sources

To be expanded into full numbered references in production. Principal sources drawn on for this draft include:

- APEC Action Plan on Rare Diseases (APEC Life Sciences Innovation Forum; endorsed by APEC Health Ministers, 2018); apec.org and rarediseasepolicy.org.
- UN General Assembly resolution on persons living with a rare disease and their families (2021); Rare Diseases International.
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