Data Generation for Rare Diseases, a tool for data-driven policymaking

A Greek Case Study

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Patient Real World Data possible uses

Clinical Trial Optimization

Identify best sites and most appropriate patients. Support protocol design

- Protocol Design & Feasibility
- On-going protocol adjustment
- Leverage RWD for study comparative arm
- Optimise country allocation
- Site & patient selection

Epidemiology Assessment

Monitoring of pathology evolution and therapeutic strategies

- Understand the natural history of disease
- Characterize patient populations & identify subgroups of interest
- Treatment pathway
- Determine the standard of care
- Identify unmet needs
- Identify suitable local comparators
- Patient flow analysis/ patient journey
- Adherence studies
- Off-label use

Drug Safety & Risk Management

Segment, analyze and assess the safety and risk/benefit of therapeutic interventions in a real-world setting

- Signal detection and assessment
- Safety Surveillance
- Vigilance
- Risk Assessment
- DUS (Drug Utilization Study)
- PASS (Post Authorization Safety Study)

HEOR/ Market Access

Demonstrate the value of medicine through evidence-based health economic evaluation and real-world outcomes for optimal pricing, reimbursement and coverage potential

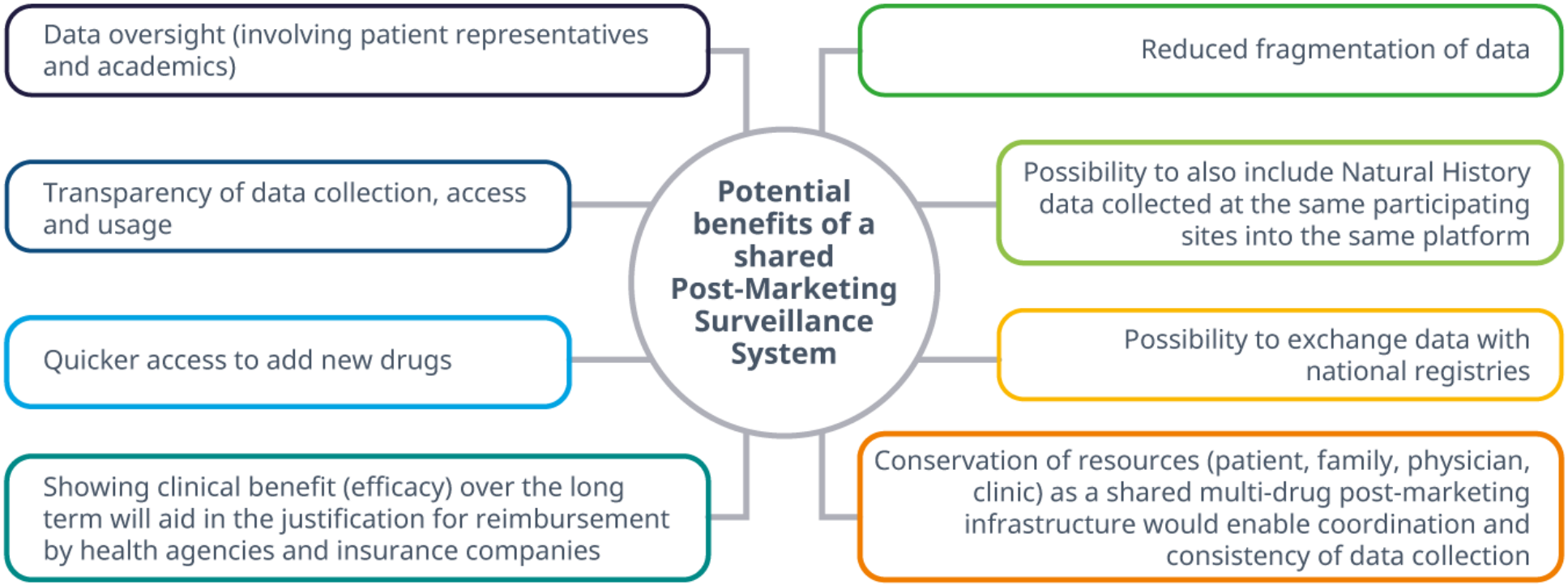
- Cost of Illness/HCRU (Health Care Resource Utilization)
- Burden of Disease
- Budget Impact
- Outcomes studies
- Comparative Effectiveness
- Compliance & Persistence
- Contract Optimization
- Target population

Commercial Analytics

Diagnose, plan,forecast and track brand performance. Size and characterize the target market from the disease and treatment pattern perspective

- Brand/launch Planning & Strategy
- Market sizing and forecasting
- Brand Diagnostics
- Brand Performance tracking/Source of business
- Split by indication
- Contract Compliance

Benefits of potential shared post-market surveillance platforms



"Regulators have expressed a preference for the development of disease-specific PMS systems over drug-specific ones, especially for rare diseases [and]..., there is an increasing recognition by industry that a shared postmarketing infrastructure for a specific disease group would facilitate and expedite PMS, while conserving resources"

Source: TREAT-NMD Neuromuscular Network website.
Report: Supporting Patients through Research Collaboration. IQVIA Institute for Human Data Science, October 2023.

This project is a collaboration between “95” Rare Alliance Greece and IQVIA

Main project leaders

IQVIA™

The main objective is to generate evidence and develop the strategy for Rare Diseases in Greece

Goal and objectives

Development of a National Action Plan for Rare Diseases, that will ensure access to optimal care and treatment for all patients living with a Rare Disease in a sustainable and resilient Health Care System

Primary objectives

- **Evidence Generation**
  - Collection and Analysis of Real-World Data about Rare Diseases in Greece based on Quantitative + Qualitative Methodology.
- **Strategy Development**
  - Propose RDs Policy Recommendations
  - Develop RDs National Action Plan Roadmap from prevention, diagnosis, care, research to access for RD in Greece

Study designed and implemented in 3 phases

Data Collection

- Desk research on RDs Prevalence
- List of Rare Diseases and prevalence in Greece

Data Analysis

- Estimate RDs economic burden
- Conduct RDs Cost analysis in Greece

Report

- Measure quality of life and patients' burden living with rare diseases in Greece

Publications

Epidemiological data

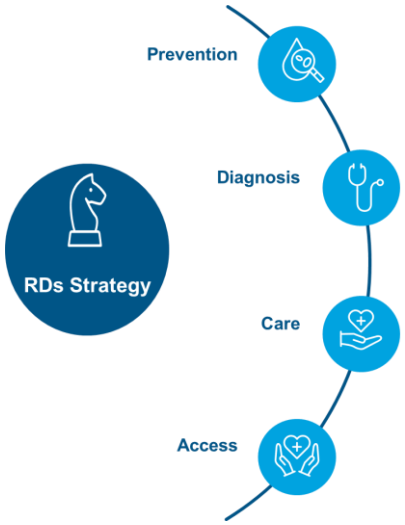
Cost of RD

Quality of Life

Outcome

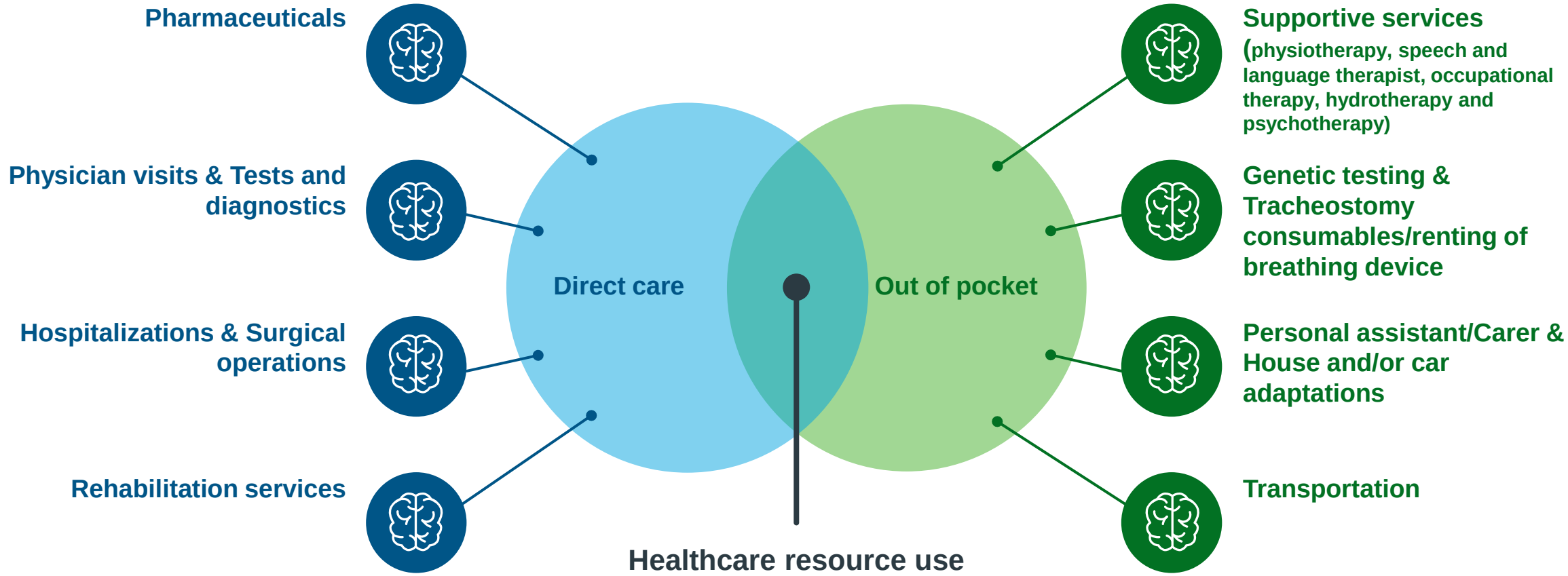
RDs National Plan

- Propose RDs Policy Recommendations
- Develop RDs National Action Plan Roadmap from prevention, diagnosis, care, research to access for RD in Greece



Data collection

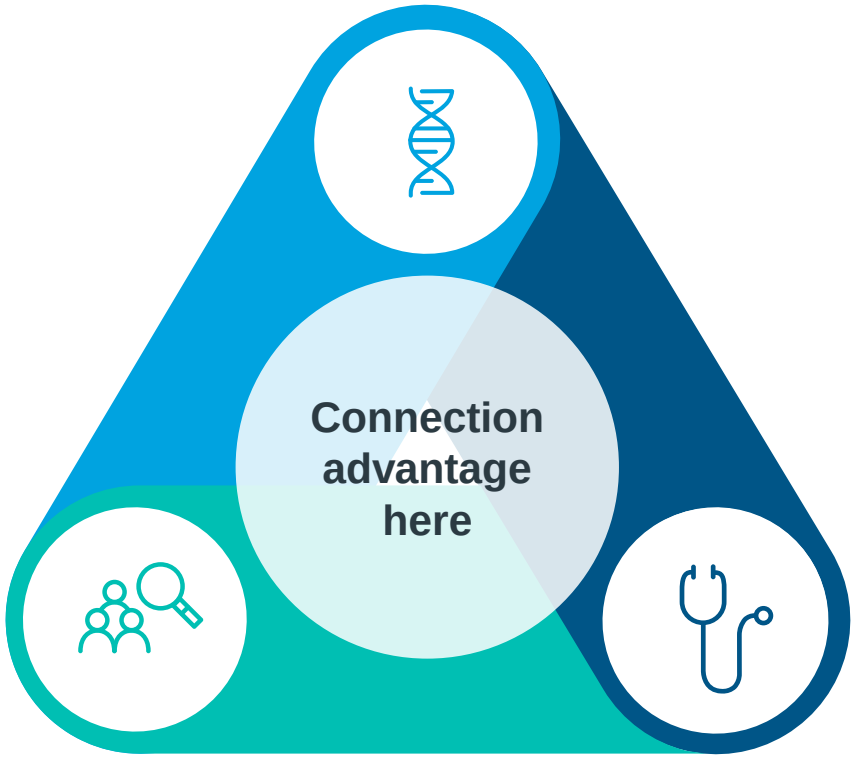
Collected and analyzed for the 1st time in Greece



Costing methodology

Pillars

Actionable
prenatal screening programs



Actionable
newborn screening programs

Actionable
early vs. late diagnosis

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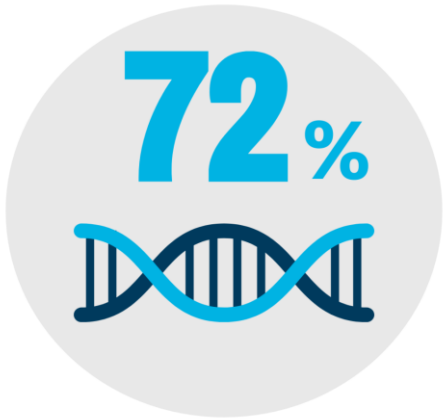
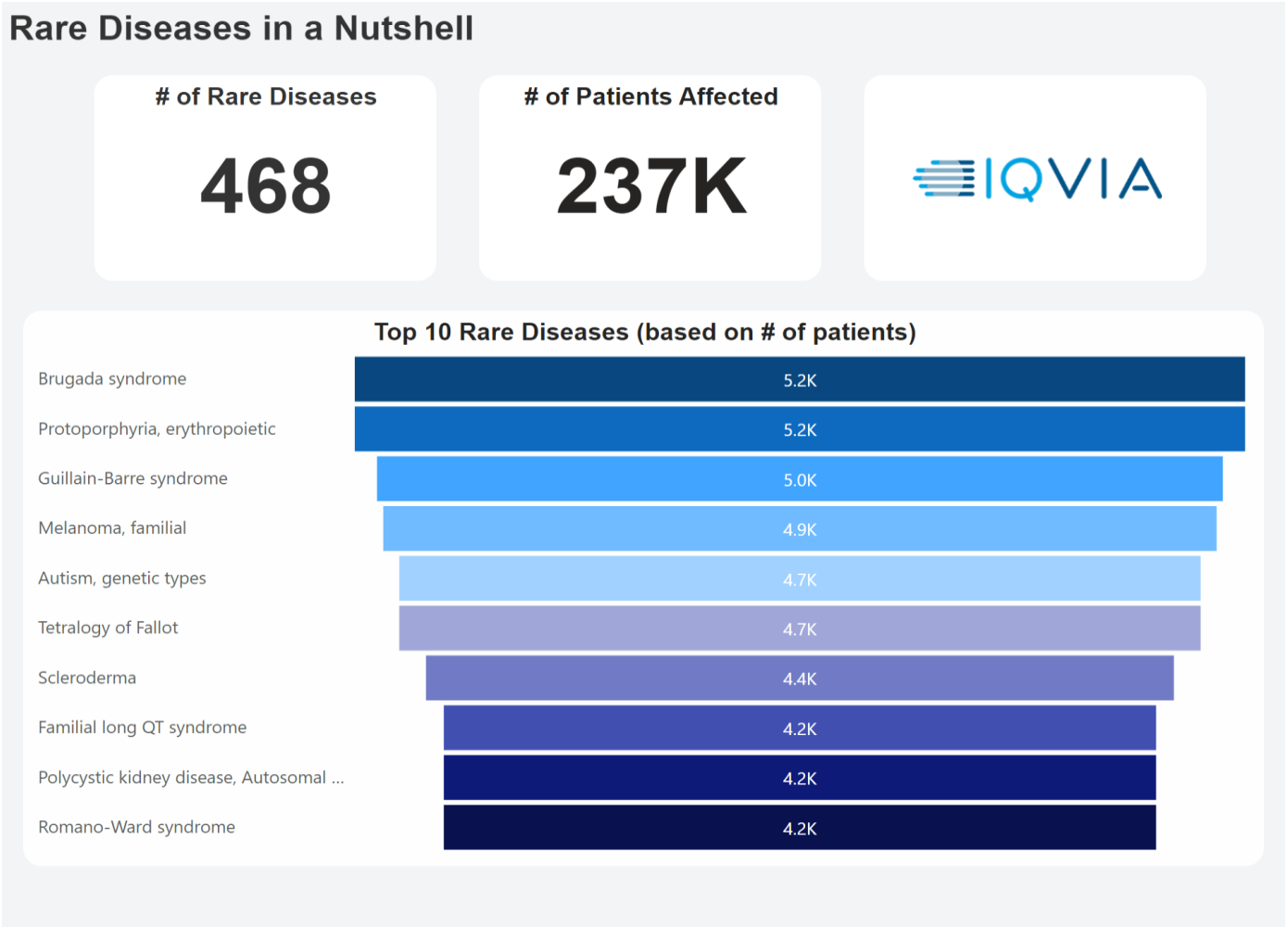
Epidemiological data

237k patients in Greece





Spinal Muscular Atrophy

Economic and humanistic burden of SMA is significantly high

SMA patients
190-348 patients
(0.003% of total population)

0.6% of the total health expenditure in Greece

SMA
- incidence of 1 in 6,000 to 1 in 10,000 live births and a carrier frequency of 1/40 to 1/60
- 2nd most common fatal autosomal recessive disorder after cystic fibrosis

€261,785

total cost (direct & indirect) per SMA patient per year (vs. €7,111/patient per year for diabetes - 10% of population)

Out of pocket costs
of the total expenditure (€20,846 on average per year/per patient) **8.0%**

Reproductive carrier screening program
Annual investment of €1,871,922 will lead to annual cost-savings equal to

€167k

€50-91m total cost annually

HRQoL
mean total score referring to SMA impact, as a chronic health condition **45.00 (max 100)**

Newborn screening program
cost-saving after the 17th year following its implementation with yearly cost-savings

€102k

Duchenne Muscular Dystrophy

Economic and humanistic burden of DMD is significantly high

DMD patients
240-522 patients
(0.005% of total population)

0.4% of the total health expenditure in Greece

DMD
- incidence of 1 in 6,000 in 10,000 live boys
- genetic disorder characterized by progressive muscle degeneration and weakness

€134,088

total cost (direct & indirect)
per DMD patient per year

€34-70m total cost annually

Out of pocket costs
of the total expenditure (€17,610 on average per year/per patient) **13.1%**

HRQoL
mean total score referring to DMD impact, as a chronic health condition

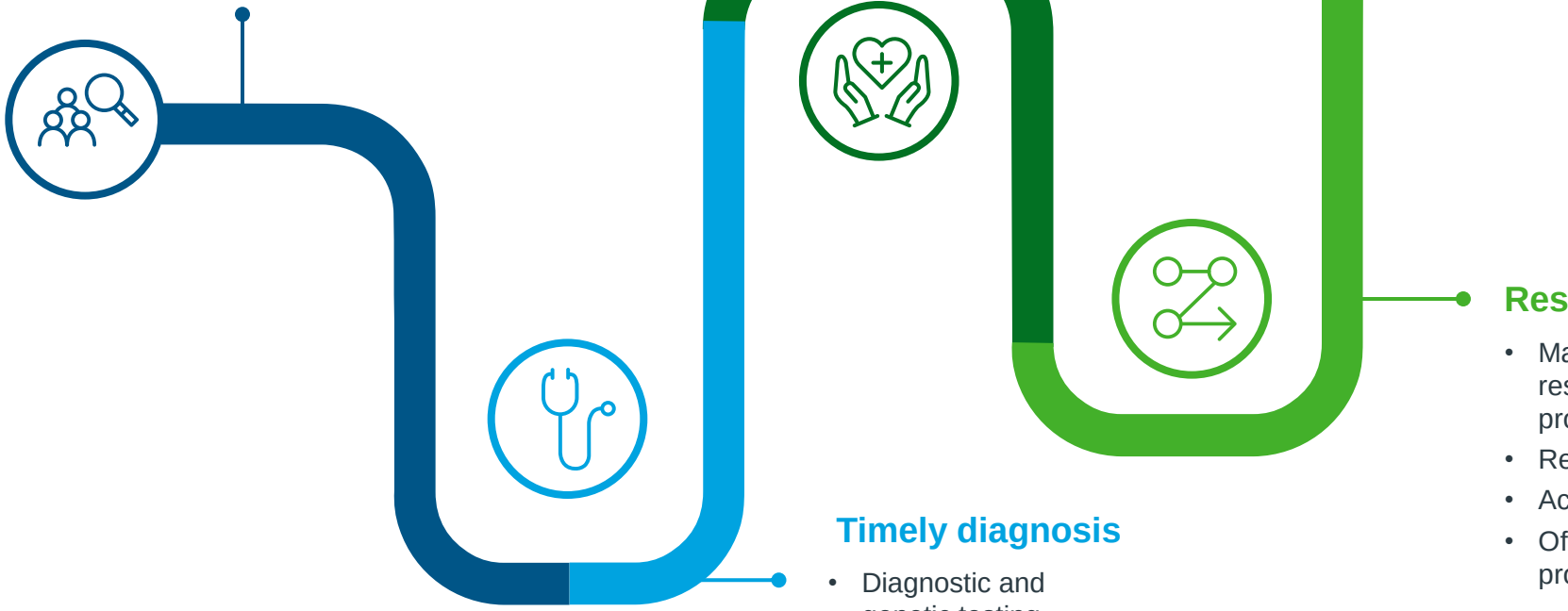
44.60 (max 100)

Comparative data

	SMA	DMD	NF	GD
Population (% of total population)	190-348 (0.003%)	240-522 (0.005%)	2,782 (0.03%)	174 (0.002%)
Annual cost per patient	€262k	€134k	€23k	€136k
Mean annual total cost (% of total health expenditure)	€50-91m (0.6%)	€34-70m (0.4%)	60m (0.4%)	24m (0.1%)
Direct cost	*92%*	*87%*	*91%*	*98%*
Mean annual out of pocket cost per patient (% of total disease cost)	€20,846 (8.0%)	€17,610 (13.1%)	€2,097 (9.1%)	€2,419 (1.8%)
Mean QoL score	45.00	44.60	60.25	34.25

Rare Diseases National plan

Roadmap from prevention, diagnosis, care, research to access



Prevention

- Develop Screening Policies
- Dedicated RD research programmes and governance of RD research funds

Timely diagnosis

- Diagnostic and genetic testing
- Ensure quality of RD diagnosis laboratory

Care-therapy

- Centers of excellence
- Timely access to optimal care
- Compassionate use programmes

Research & education

- Mapping of existing research resources, infrastructures and programmes for RDs
- Registries
- Access to treatments
- Off label use of medicinal products

Thank you!

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