

Prenatal and neonatal screening programs as a tool for a sustainable Healthcare system

Data Generation for Decision Making for RDs in Greece

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3ο Διεθνές Συνέδριο για τις Σπάνιες Παθήσεις: Ελληνικό Παράρτημα,
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Strategic collaboration between “95” Rare Alliance Greece and IQVIA

Main project leaders



The main objectives are to generate evidence and develop the strategy for Rare Diseases in Greece

Goal and objectives



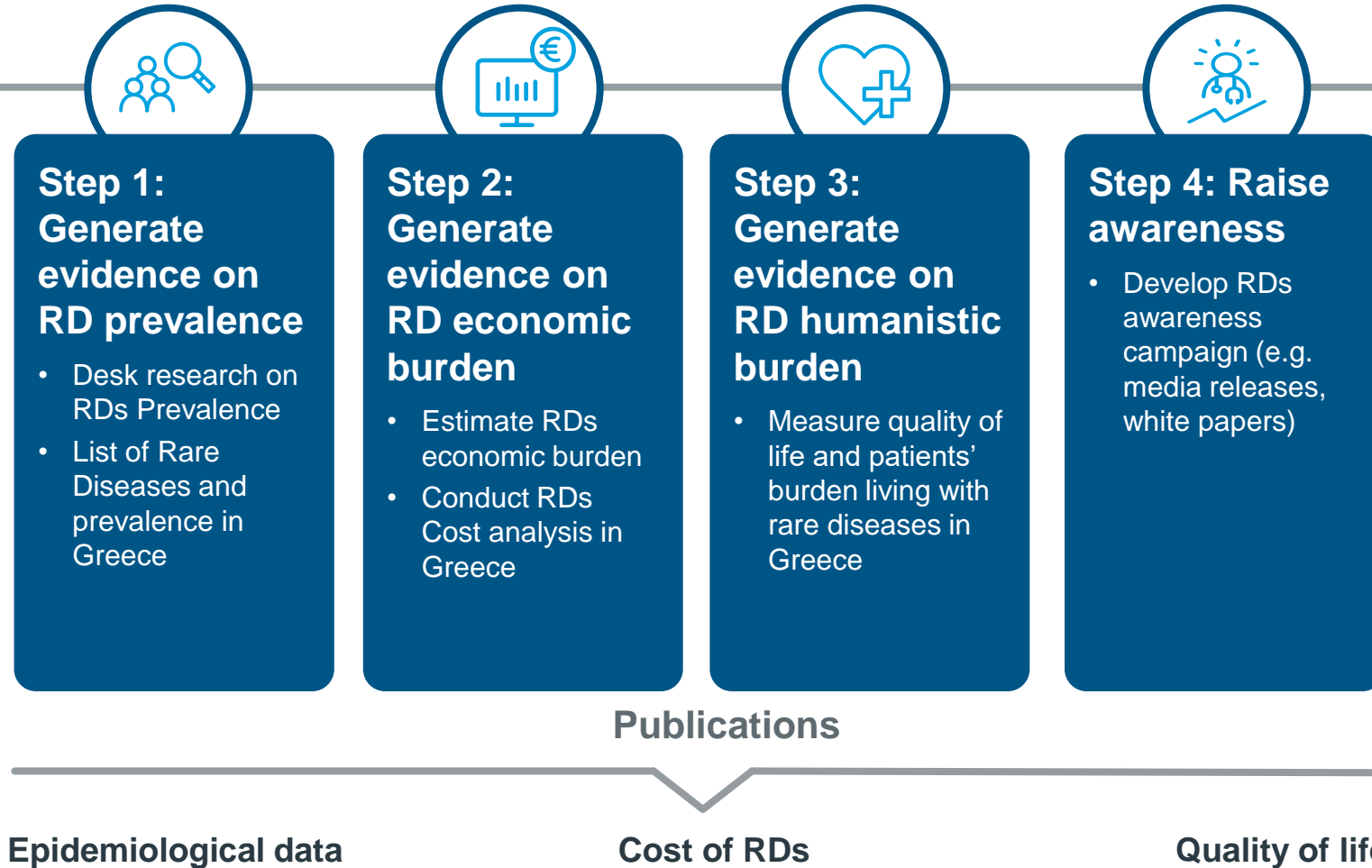
Development of a National Action Plan for Rare Diseases, that will ensure access to optimal care and treatment for all patients living with a Rare Disease in a sustainable and resilient Health Care System

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

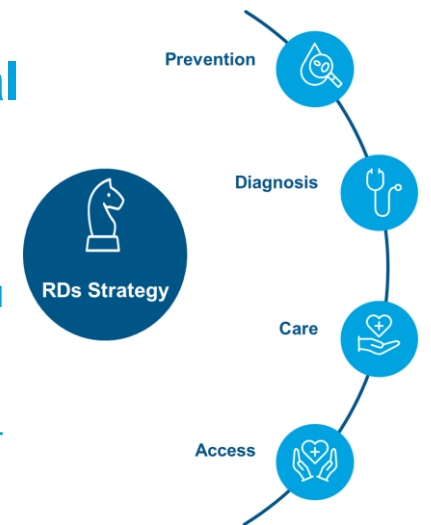
Approach

Consisting of 5 steps



RDs National Plan

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



Spinal Muscular Atrophy

Economic and humanistic burden of SMA is significantly high

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



1.2% **cost of management of SMA**
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

€514,654



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

Out of pocket costs
of the total expenditure (€17k on
average per year/per patient) **6.5%**

**Reproductive carrier screening
program**
~ €2-2,5m avoided costs resulting to
cost-savings of

€140-708k

€98-179m total cost over a
two-year timeframe

HRQoL
of parents/ carers
scores in the majority
of the scales falling
below **50 (max 100)**



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

€106k

Duchenne Muscular Dystrophy

Economic and humanistic burden of DMD is significantly high

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



0.8% **cost of management of DMD**
of the total health expenditure
in Greece



DMD

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

€254,053

total cost (direct & indirect)
per DMD patient over a two-
year time period



Out of pocket costs

of the total expenditure (€11k on
average per year/per patient)

8.7%

**Reproductive carrier screening
program**
avoided cost

HRQoL

of parents/ carers
scores in the majority
of the scales falling
below

50 (max 100)



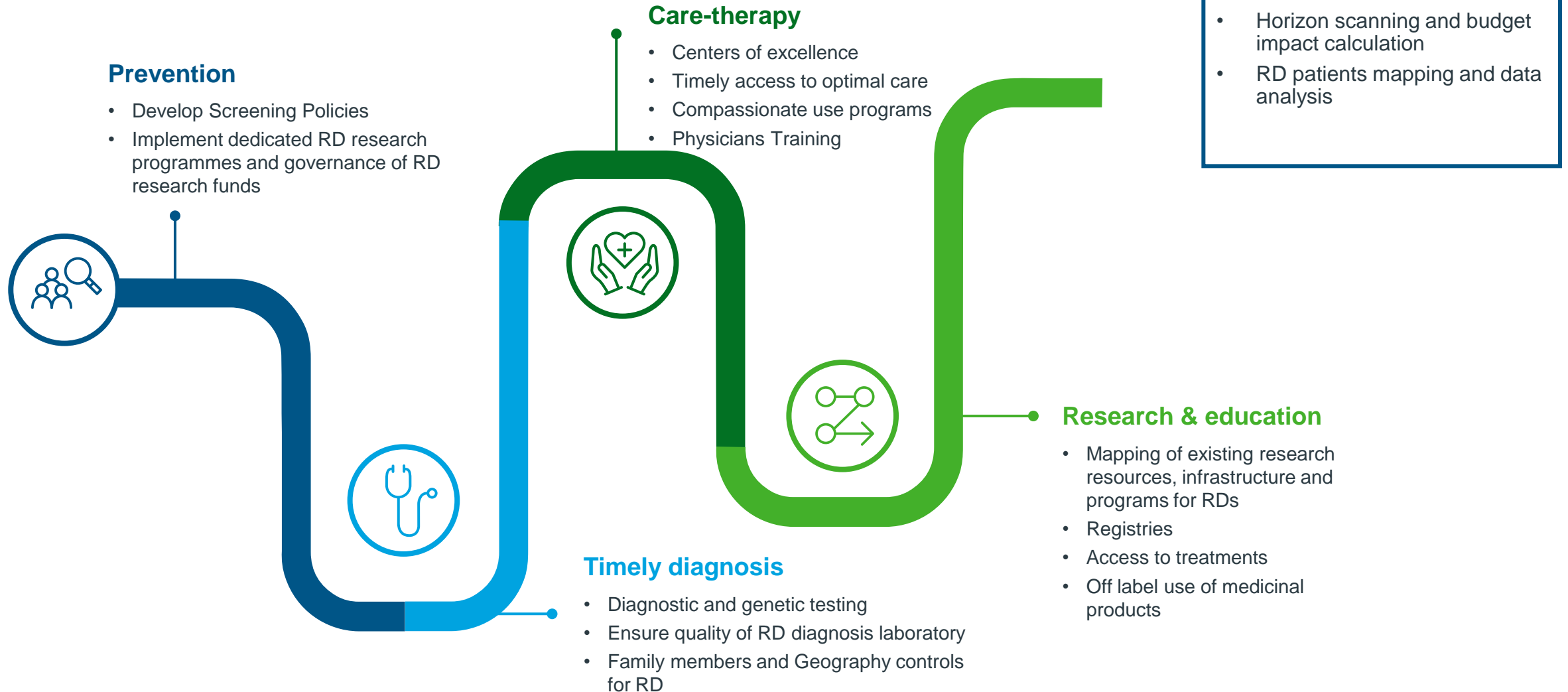
€750k

€61-133m

total cost over a
two-year timeframe

Rare Diseases National plan

Roadmap from prevention, diagnosis, care, research to access





Thank you!