

# *EUROPEAN REFERENCE NETWORKS GUIDELINES, KNOWLEDGE GENERATION AND TRAINING*

*MAURIZIO SCARPA*

*COORDINATOR,*

*EUROPEAN REFERENCE NETWORK*

*HEREDITARY METABOLIC DISEASES, MetabERN*

*PAST-CHAIR,*

*EUROPEAN REFERENCE NETWORK*

*COORDINATORS GROUP, ERN-CG*



According to patients surveyed, it takes:



on average **7.6 years in the US**



on average **5.6 years in the UK**  
for a patient with a rare disease to  
receive a proper diagnosis

According to patient/caregiver respondents, in order to  
get a proper diagnosis, a patient typically visits up to

**8 physicians: 4 primary care and 4 specialists**



and receives **2** to **3** misdiagnoses



**The majority** of physicians reported it is more difficult to address the needs of a rare disease patient in a typical office visit.



**Nearly all** physicians stated more office visits are required to diagnose a rare disease patient.



**The majority** of physicians said it takes more office visits to adequately address symptoms.



**60%**  
in the US



**50%**  
in the UK

**Around half** of patients with a rare disease and their caregivers stated they received conflicting information from different healthcare professionals about treatment options.



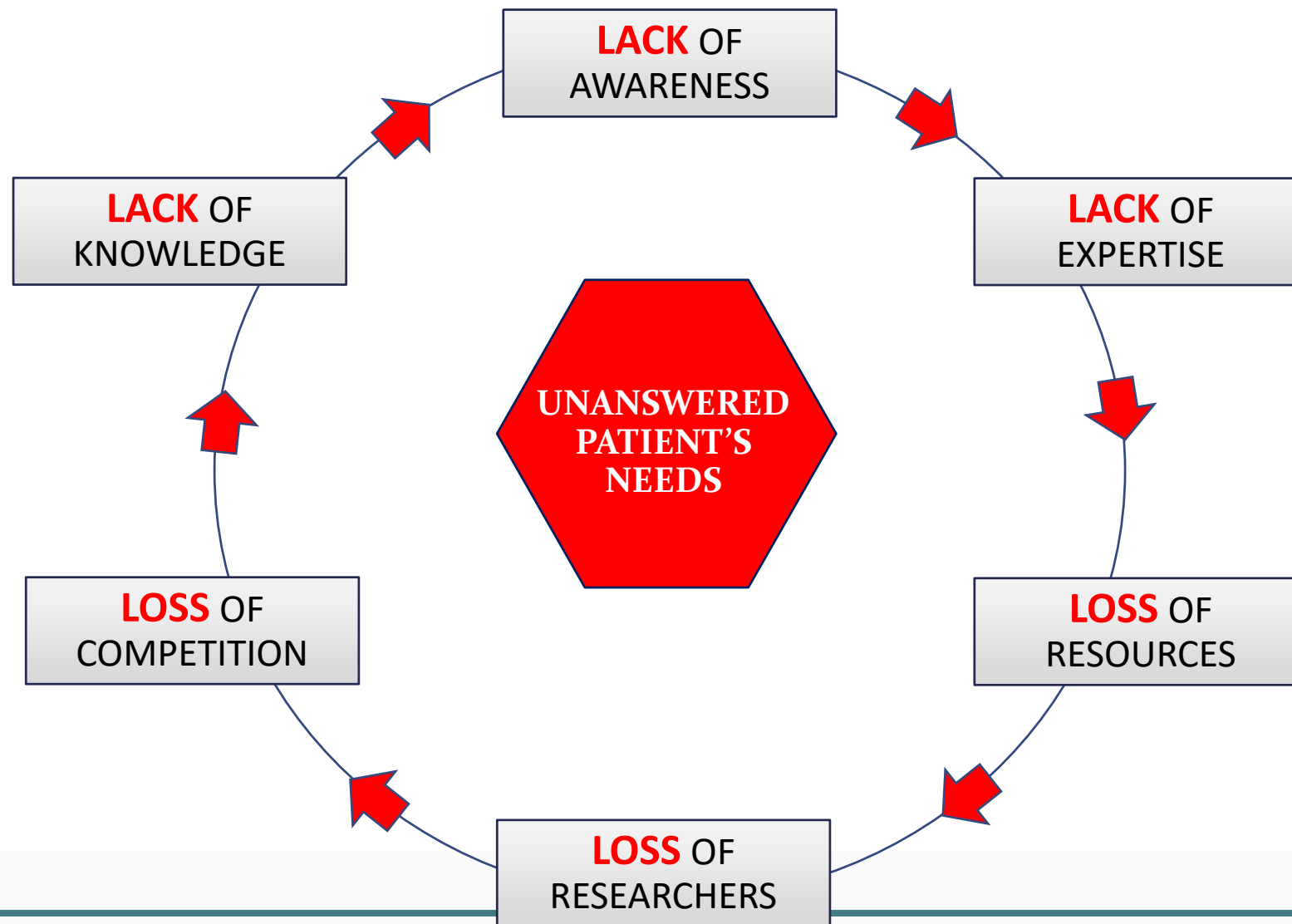
**67%**  
in the US



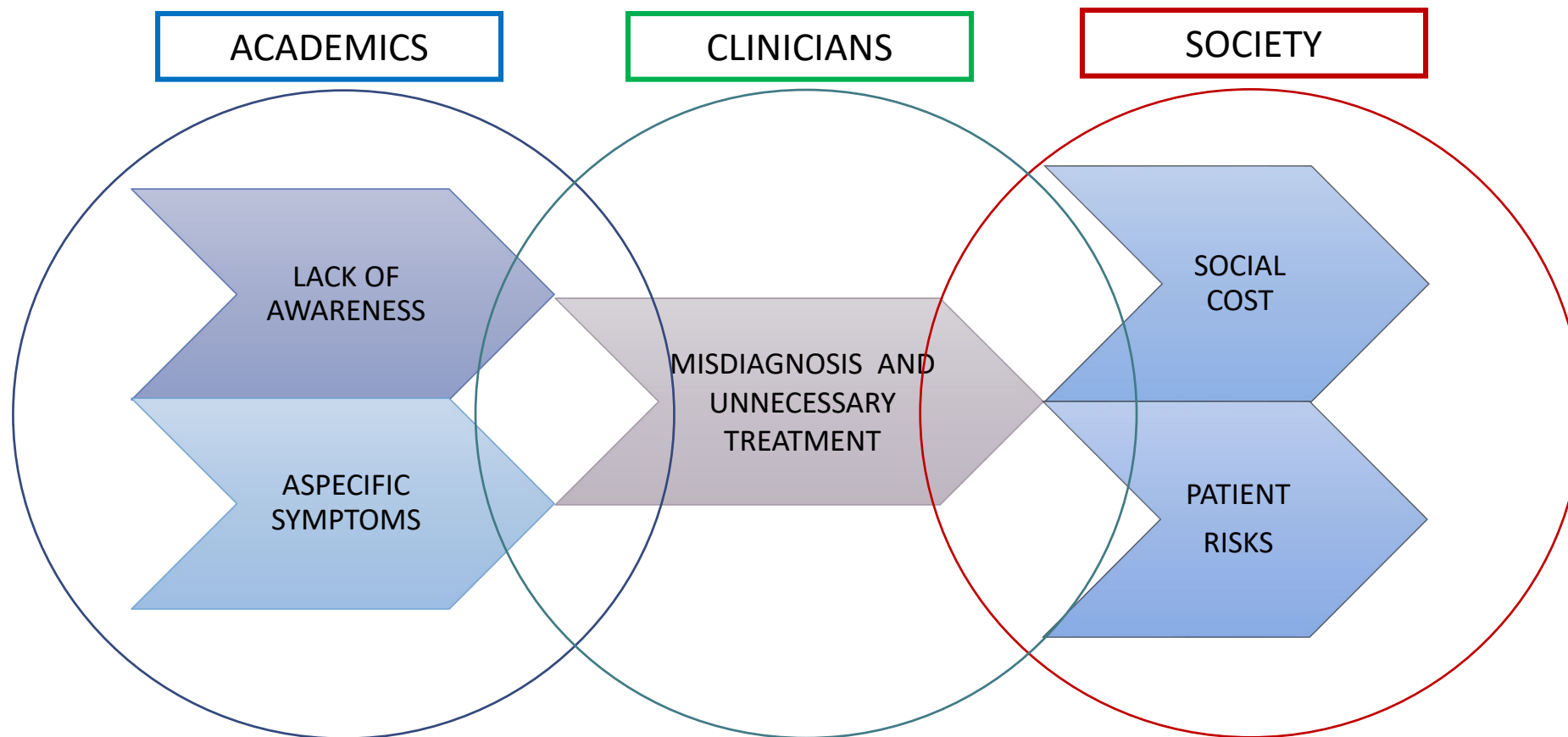
**62%**  
in the UK

**More than half** of patients and caregivers stated they needed to provide their healthcare professionals with information on their rare disease.

# THE RARE DISEASES VICIUS CIRCLE



# THE EFFECT CHAIN OF LOSS OF EXPERTISE



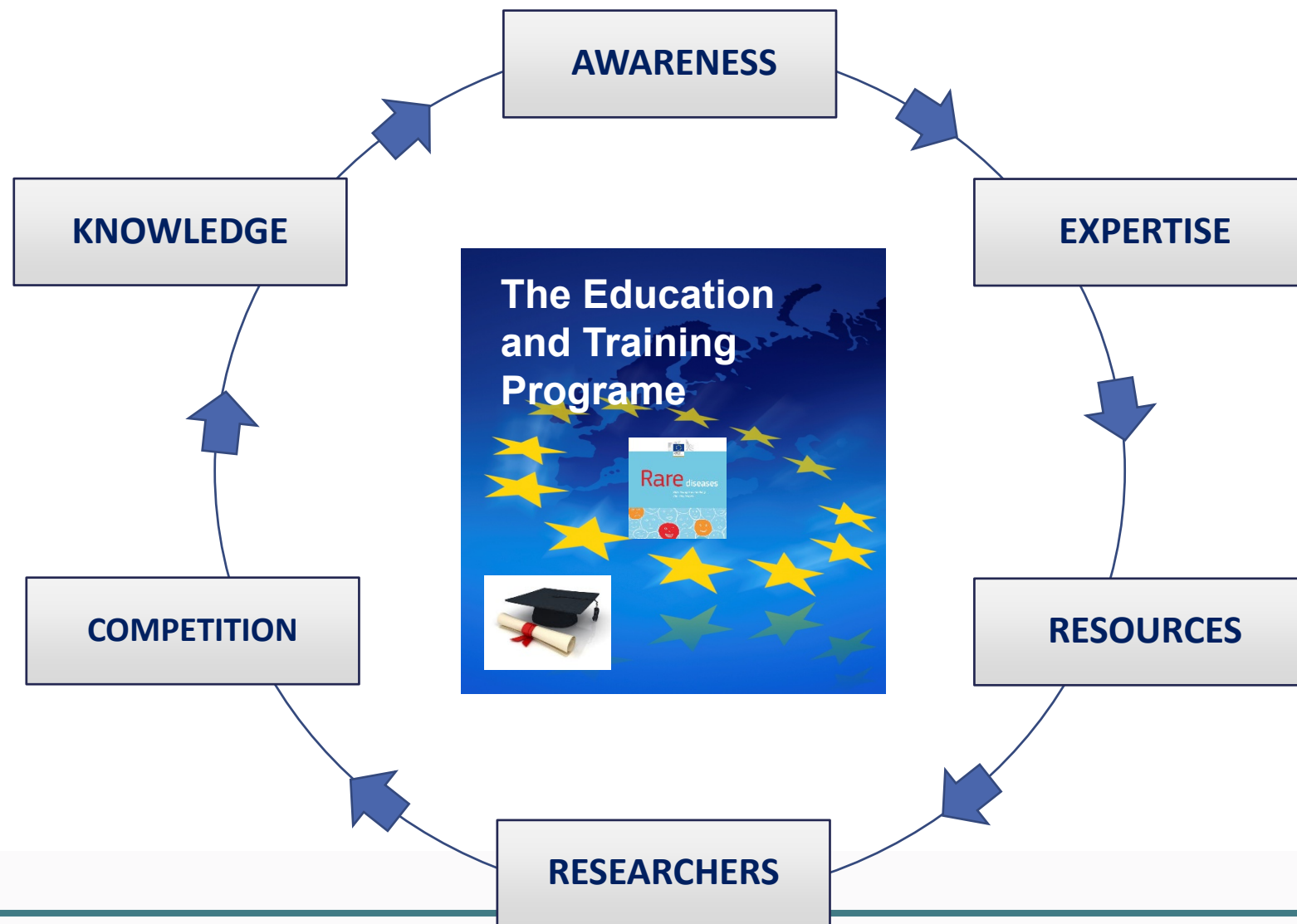
# Education and Training Program

## Protect the future

<p><i>Address the gaps in knowledge of rare disorders</i></p>	<p><i>Cross-sector cooperation to create synergies and better share available expertise</i></p>	<p><i>Reconcile societal needs of patients, curricula of healthcare professionals and biomedical scientists, employment opportunities and research dynamism</i></p>
---------------------------------------------------------------	-------------------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------------------------------------------------------------------------------------

*European Reference Network, Stakeholders, BoMS, EC*

# THE ANSWER TO PATIENT'S NEED





## *PRIORITY ELEMENTS TO BE DISCUSSED IN THE WG*

- WHITE BOOK ON KNOWLEDGE GENERATION IN THE ERNs
- IDENTIFICATION OF COMMON PLATFORMs FOR THE PRODUCTION OF TRAINING MATERIAL
- IDENTIFICATION OF COMMON PLATFORMs FOR E-LEARNING
- SETTING UP A PROOF OF CONCEPT PROGRAM FOR THE INTRODUCTION OF RARE DISEASES IN THE MEDICAL CV
- LAUNCH OF INTER-ERN PILOT PROGRAMS FOR EDUCATIONAL MOBILITY OF POSTGRADUATED PROFESSIONALS
- INTRODUCTION OF KNOWLEDGE GENERATION IN THE RARE DISEASE PLANS