



European
Reference
Network

for rare or low prevalence
complex diseases

 Network
Neuromuscular
Diseases (ERN EURO-NMD)



EpiCARE

Neurological Diseases
(ERN-RND)

7th ERN EURO-NMD

ANNUAL MEETING

Molecular therapeutic board Neurological Channelopathies

21st – 23rd February 2024

Dr. med. Robert Lauerer-Braun
Department of Neurology and Epileptology
University Hospital Tübingen



Universitätsklinikum
Tübingen

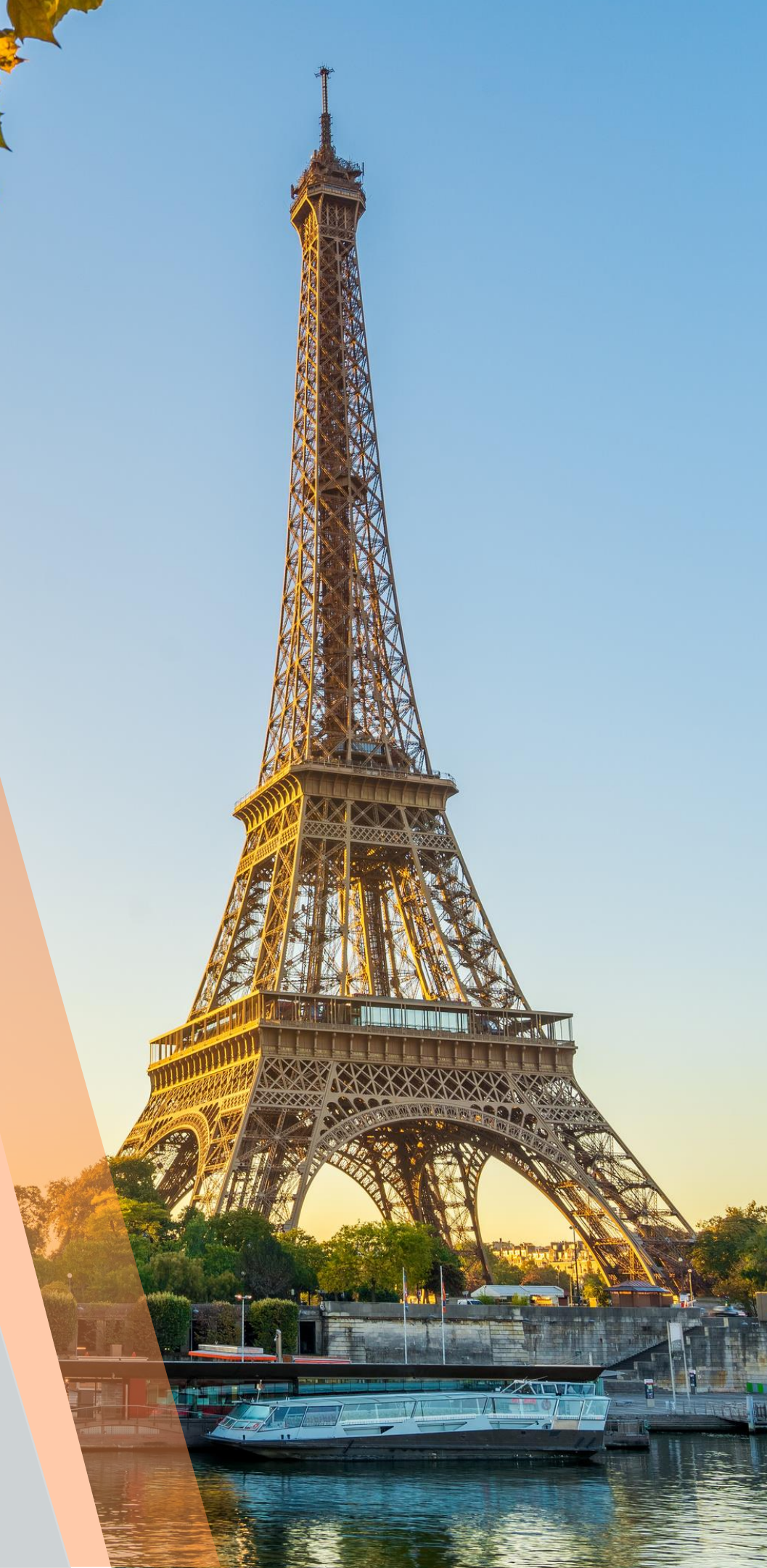


Hertie Institute
for Clinical Brain Research

EBERHARD KARLS
UNIVERSITÄT
TÜBINGEN



Funded by
the European Union

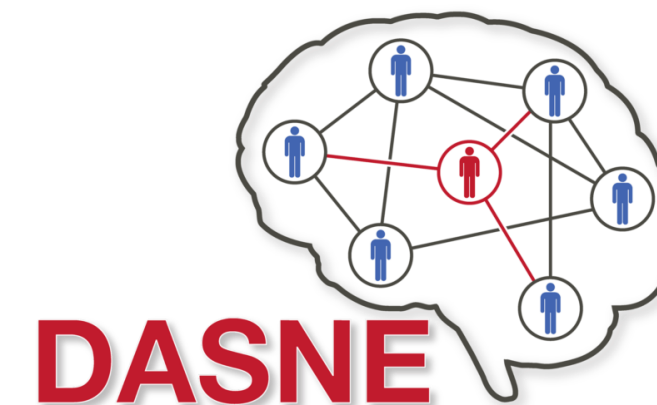


Idea of a Molecular Therapeutic Board

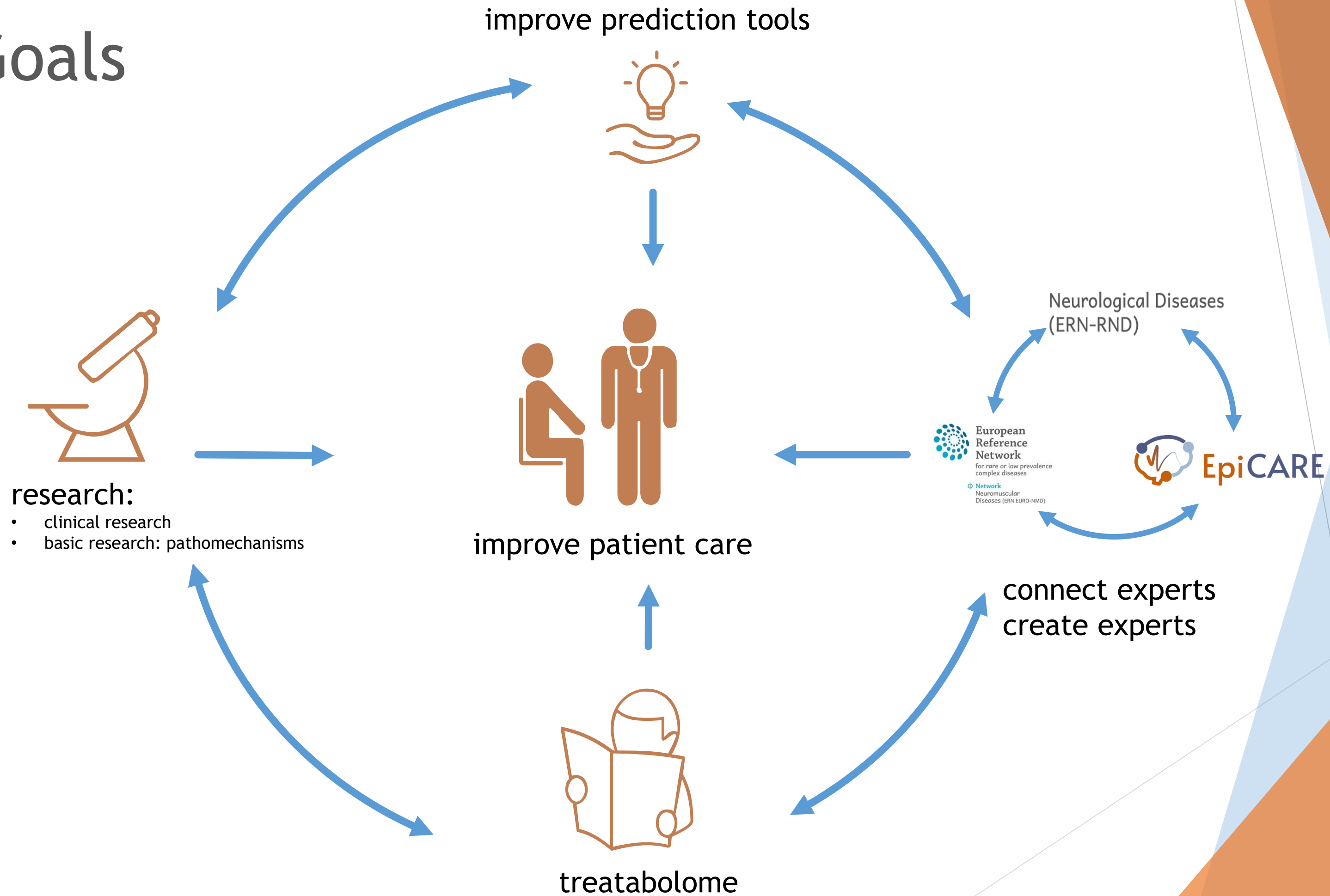


Prof. Holger Lerche

- ▶ ion channelopathies include all kinds of neurological disorders
 - ▶ neuromuscular diseases
 - ▶ movement disorders
 - ▶ epilepsy
 - ▶ ...
- ▶ ion channels are common drug targets
 - ▶ can be used for drug repurposing
- ▶ molecular therapeutic board: bring experts & knowledge together
- ▶ established format in DASNE: German academy for rare neurological disorders
- ▶ online meeting via Microsoft Teams 1h approx. every 3-4 months

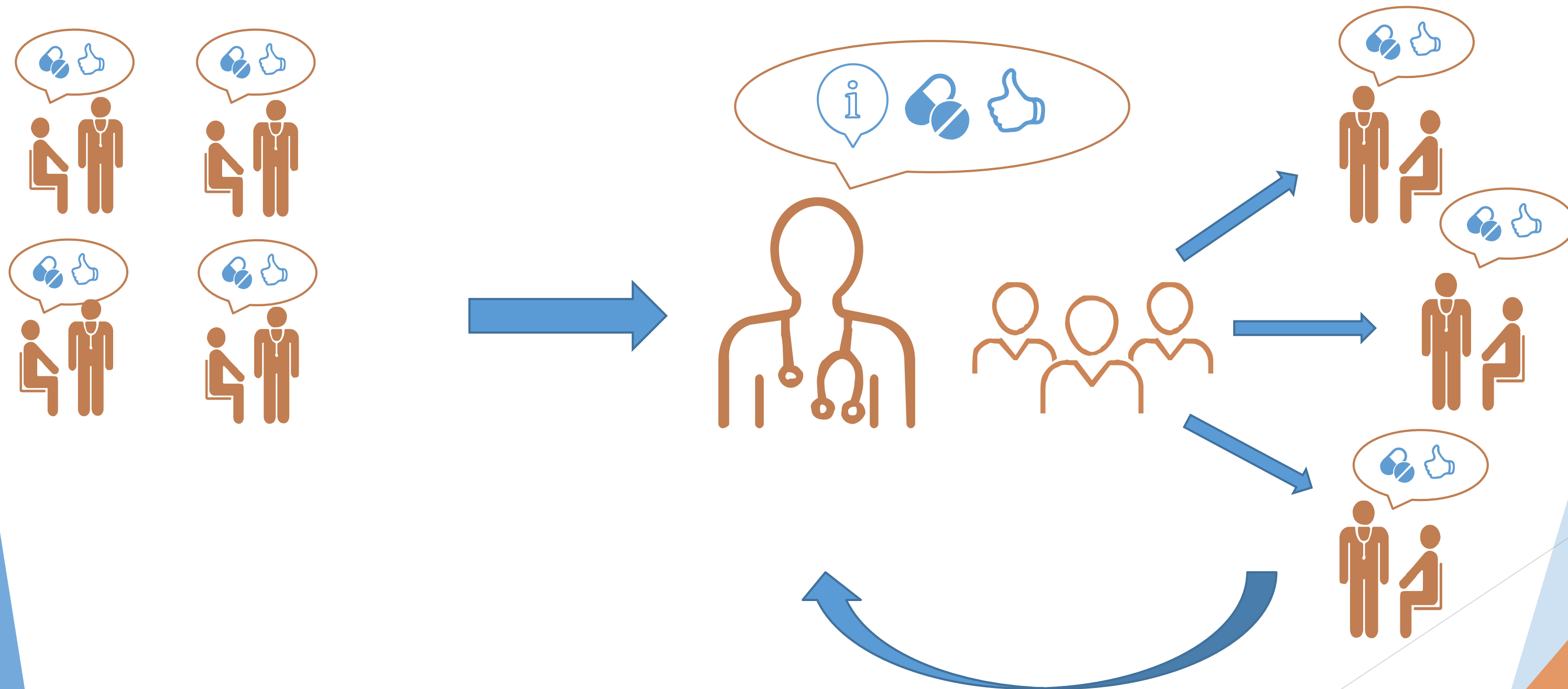


Goals



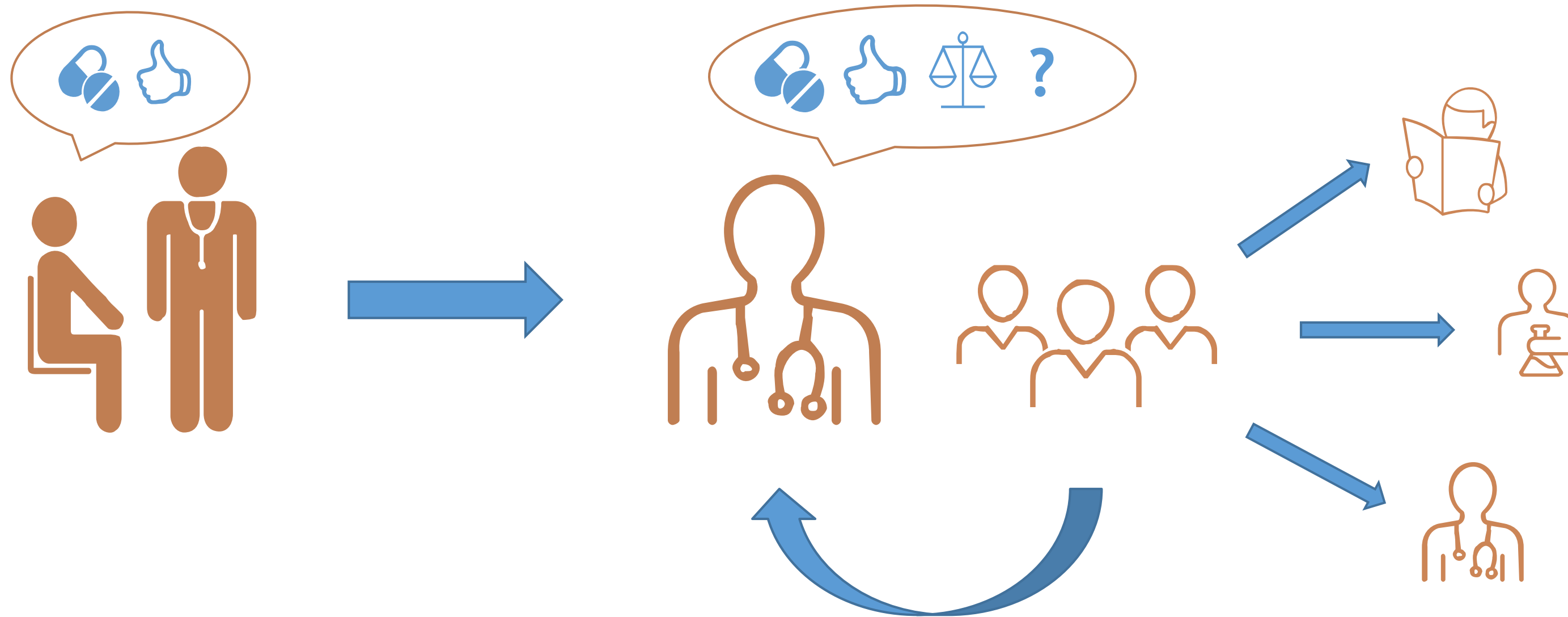
3 Ways of presentations:

1- known and tested therapies



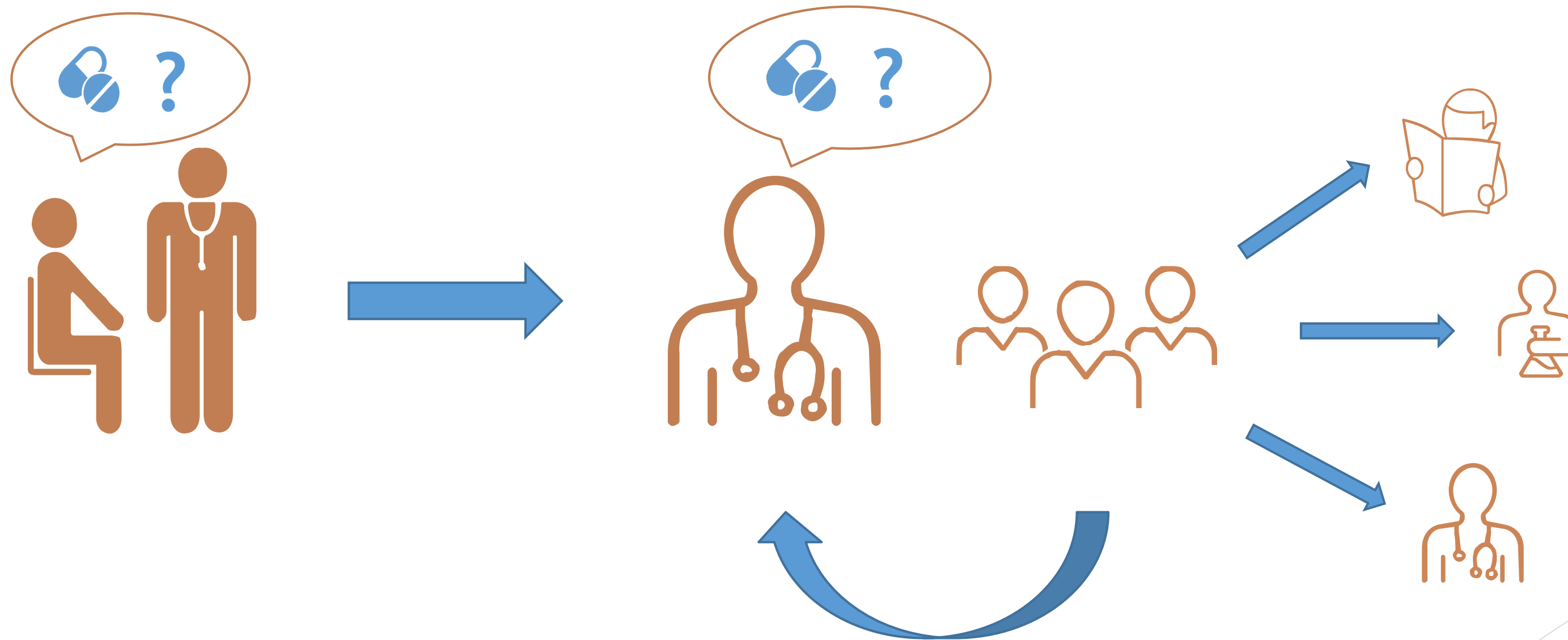
3 Ways of presentations:

2- Response to specific drug in otherwise pharmacoresistant patient



3 Ways of presentations:

3- Known genetic defect - find specific therapy



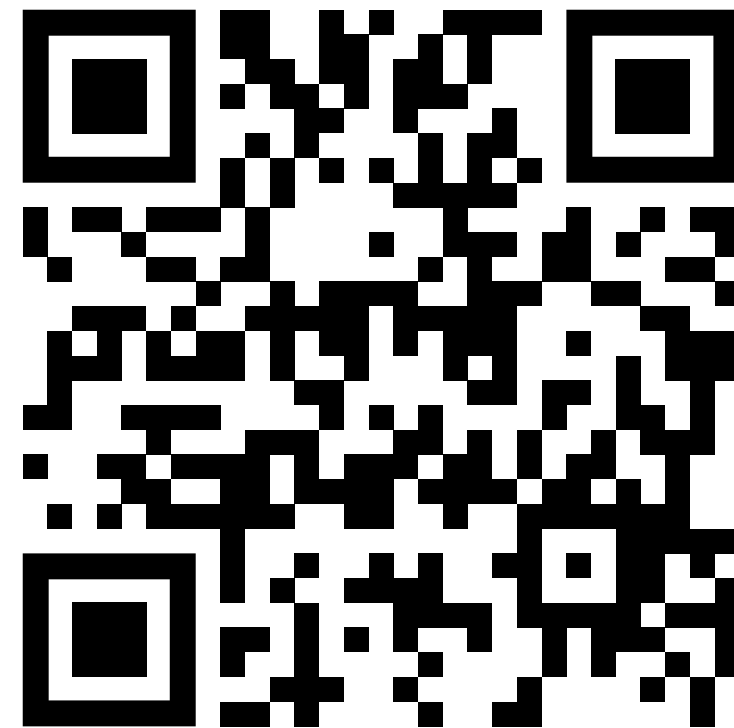
Boards so far

- ▶ 02/2023:
 - ▶ Case of *CACNA1A*-associated DEE
 - ▶ Case of response to clavulanic acid in a patient with *KCNH1* GOF associated DEE
- ▶ 07/2023:
 - ▶ *KCNA1* gain of function epileptic encephalopathy treated with 4-aminopyridine
 - ▶ *KCNA2* gain of function epileptic encephalopathy treated with 4-aminopyridine
- ▶ 10/2023:
 - ▶ Presentation of the treatabolome project
 - ▶ Targeted therapies for skeletal muscle ion channelopathies
- ▶ 01/2024:
 - ▶ Case of *CACNA1A*-associated hemiplegic migraine

Thank you!

Next board on 15th May @ 12.00 CET Submit your cases!

<https://form.jotform.com/232903437636358>



Join the board:

Nicola Openshaw-Lawrence: n-openshaw.lawrence@epi-care.eu
Robert Lauerer-Braun: robert.lauerer@med.uni-tuebingen.de