European Survey to explore the impact of epileptic encephalopathy KCNQ2 in the daily lives of affected patients.

An iniziative of:

- EUROPEAN KCNQ2 ASSOCIATION
- KCNQ2 Einblick, Beratung, Netwerk Germany
- ASOCIACION KCNQ2 ESPANA
- KCNQ2 FRANCE
- DRAVET ITALIA ONLUS

With the support of CREP Centro Ricerca Epilessie in età Pediatriche - Verona

Isabella Brambilla

DRAVET ITALIA Onlus
Associazione Italiana Sindrome di Dravet Onlus

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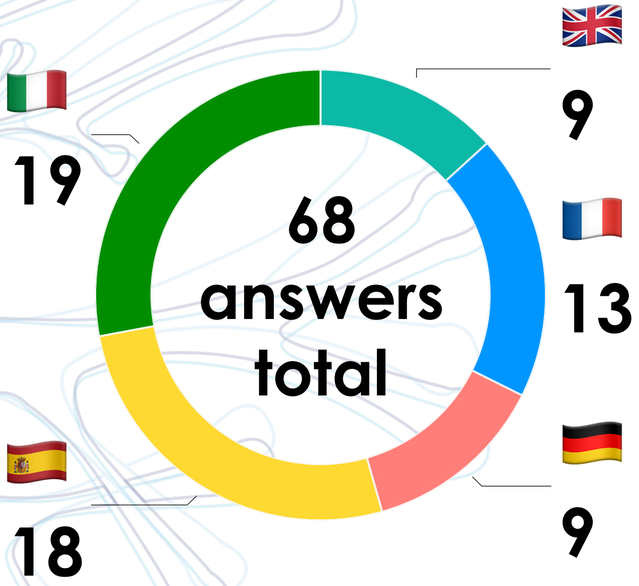
Preliminary Data Survey KCNQ2

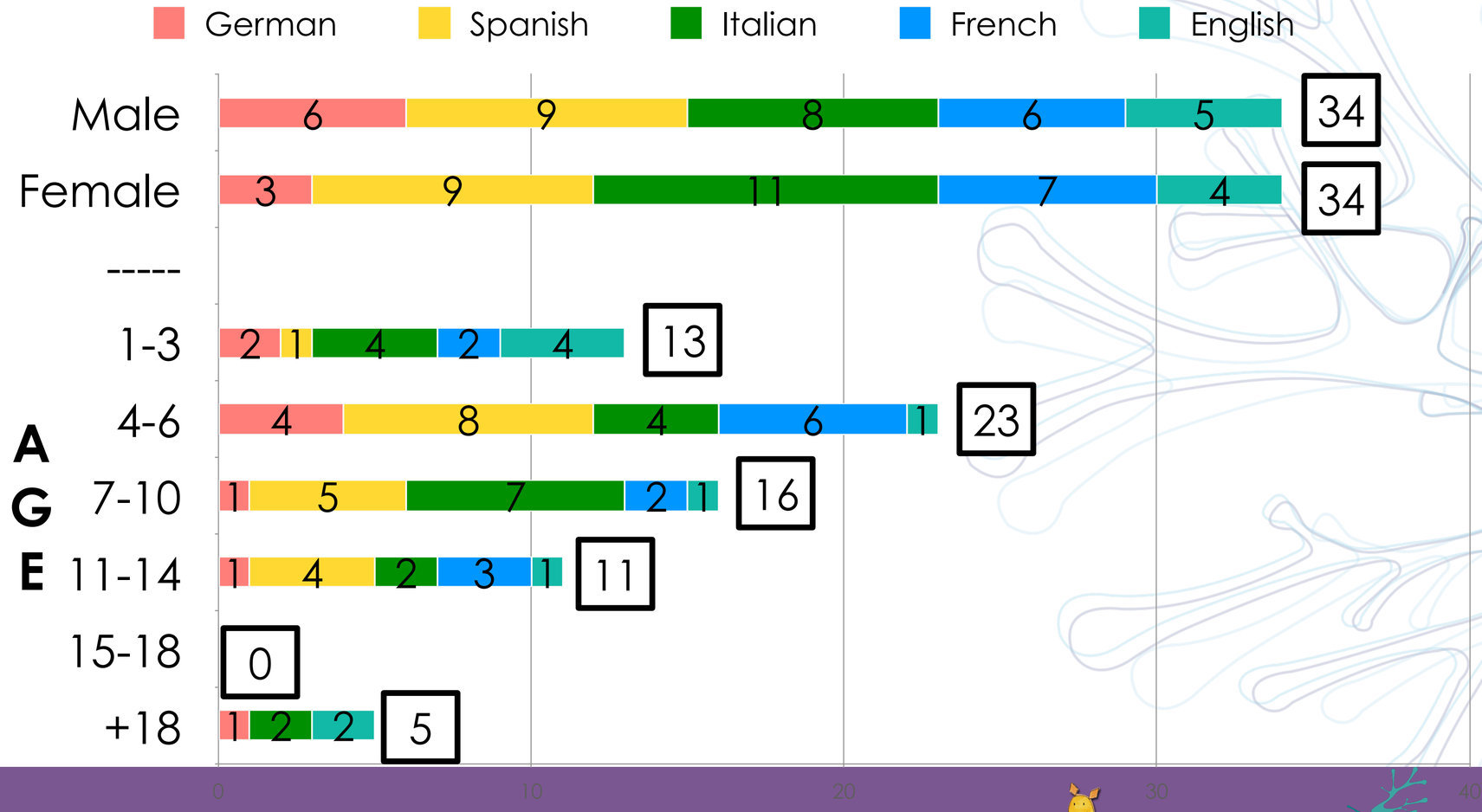
Survey translate in 5 languages ad disseminate in all word by the national associations.

Italian Version: divided in 2 times (2020 December 20 to 2021 January 10 and 2021 February 10 to March 30.)

English French, German and Spanish Version opened from 2021 February 10 to March 30.

Preliminary Data Survey KCNQ2





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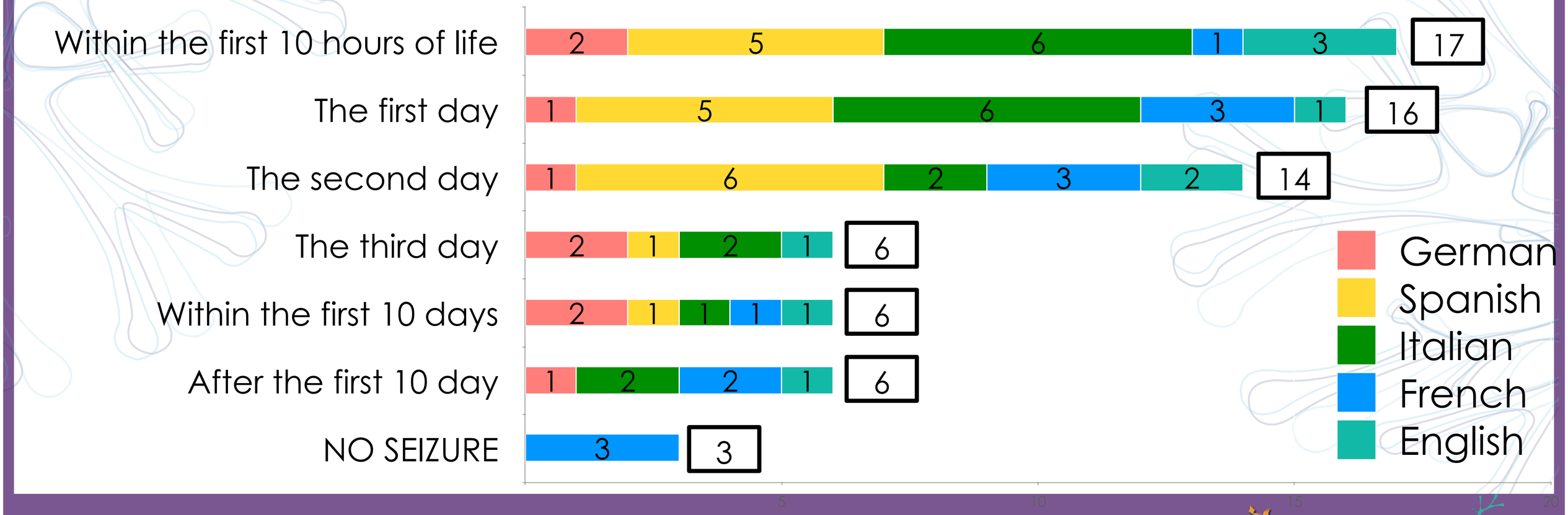
Preliminary Data Survey KCNQ2

Dr. Tommaso Lo Barco & Prof. Francesca Darra

What we already know about KCNQ2 Encephalopathy:

- It's a Developmental and Epileptic Encephalopathy with seizures starting in first days of life

SEIZURES 65/68
ONSET AGE



Dr. Tommaso Lo Barco & Prof. Francesca Darra

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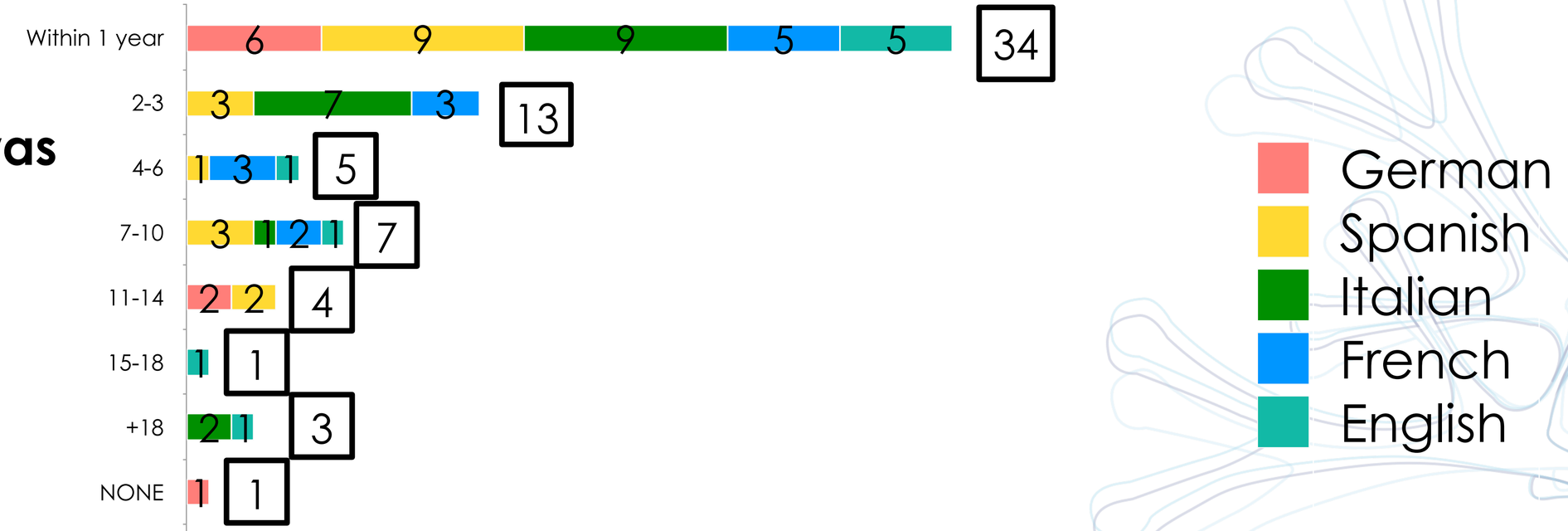
What we already know about KCNQ2 Encephalopathy:

- Mutation is almost always *de novo*

At what age was the genetic diagnosis performed?



Have genetic investigations been extended / carried out to parents as well?

Dr. Tommaso Lo Barco & Prof. Francesca Darra

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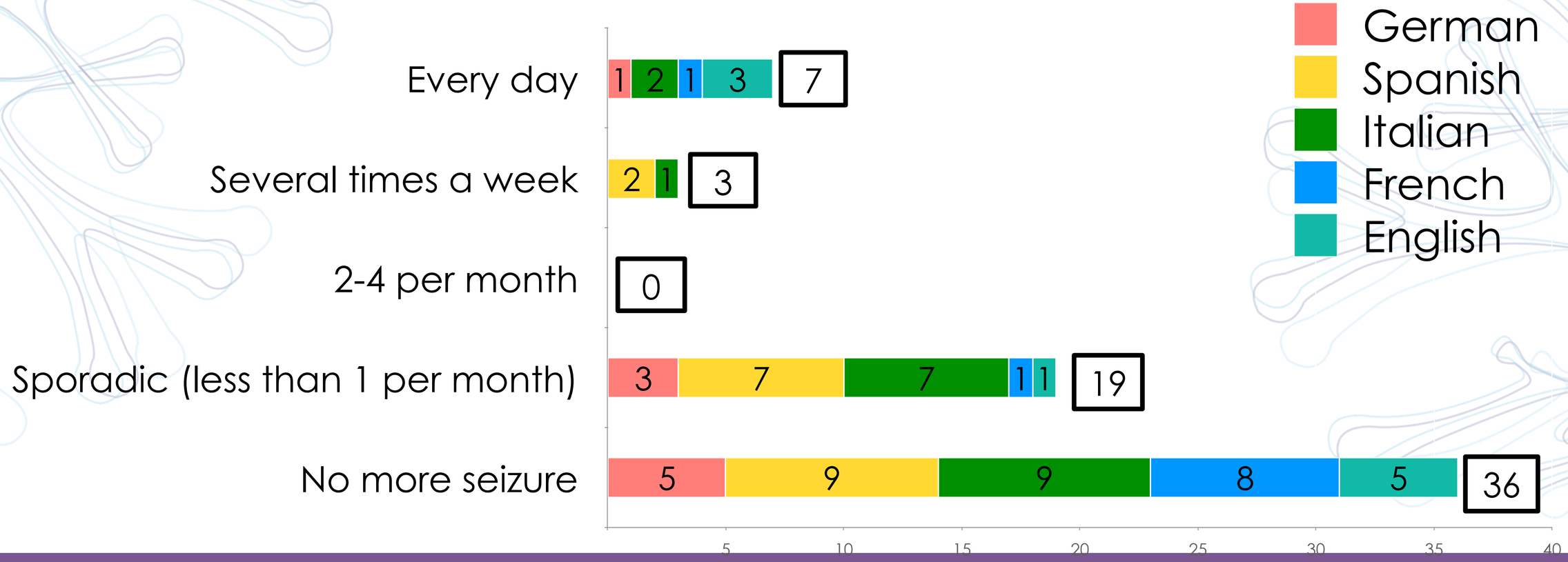
What we already know about KCNQ2 Encephalopathy:

- Seizures usually stop during first 4 years of life

MEDIAN AGE 6

How often do seizures occur today?
In 65 pts with seizures



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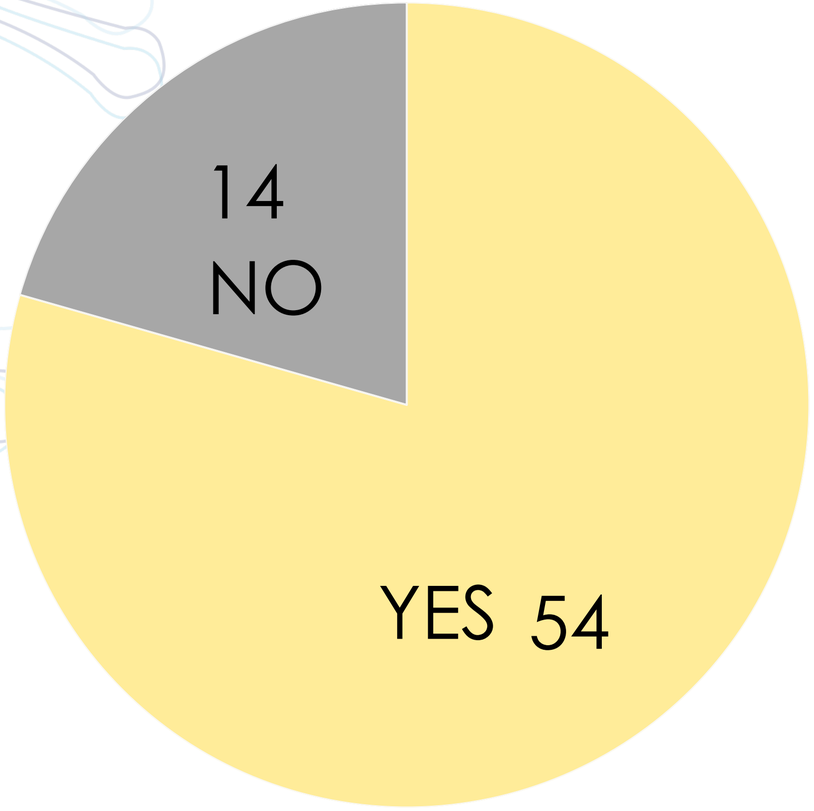
Comorbidities variously associated
54/68 pts

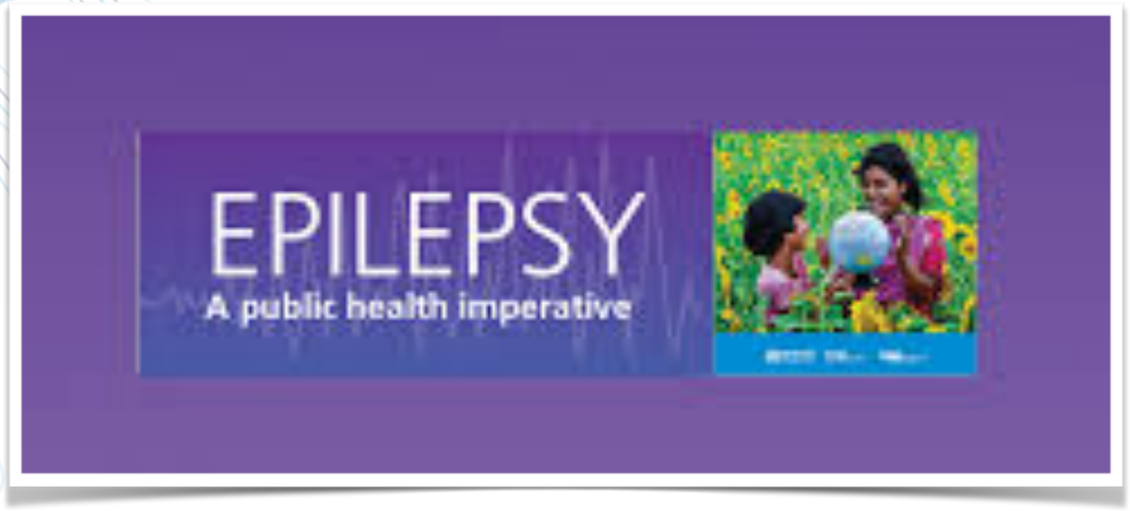


- Gastrointestinal complications
- Orthopedic complication
- Urinary problems
- Breathing problems
- Dermatological problems
- Endocrinological problems

In conclusion..

- This is the largest family survey performed on patients with KCNQ2 encephalopathy.
- It represents a unique chance to collect information regarding daylife of individuals affected by this rare condition.
- By analysing acquired data in different groups of age, we can try and delineate natural history (epilepsy trend, neurological outcome, personal autonomies)
- This would help to:
  - Individuate possible subgroups of patients with worst/better outcome
  - Define treatment outcomes (rehabilitation/pharmacological/…gene therapy?) other than seizures
  - Raise the attention on the family burden.

Thanks for the attention



World Health Organization

SEVENTY-THIRD WORLD HEALTH ASSEMBLY
Agenda item 11.6

A73/A/CONF./2
9 November 2020

Global Actions on epilepsy and other neurological disorders

Draft resolution proposed by Belarus, Bhutan, China, Colombia, Eswatini, the European Union and its Member States, Guyana, Iceland, Jamaica, Philippines, Russian Federation