Dr Caterina Lo Rizzo - SIENA
IQSEC2-related phenotype

WE JOIN CALL!

Call for collaborative clinical research on developmental disorders

These calls for collaborative projects typically aim to rapidly build up consistent clinical series for rare monogenic disorders, in order to better delineate the clinical spectrum and natural history of recently identified entities in the field of ITHACA. The call will be disseminated to the mailing list of ITHACA and will be posted on the web site.

- Short title: ***IQSEC2*-related phenotype**
- Targeted gene(s)/phenotype under study (to be quoted in the newsletter): ***IQSEC2***
- Summary (1000 letters max): ***IQSEC2*** **is a new gene related to an X-linked intellectual disability, characterized by a highly variable phenotype partially overlapping Rett syndrome. To date the mechanisms underlying this broad clinical variability are not fully understood both in male and female patients. The aim of this study is to collect clinical and molecular data of patients harbouring a mutation in *IQSEC2* gene in order to better define the clinical wide spectrum of *IQSEC2* gene related phenotype.**
- Coordinating clinician: **Prof. Alessandra Renieri, MD, PhD**
- Institution (dept, hospital, City): **U.O.C. Genetica Medica, Università di Siena – Dipartimento di Biotecnologie Mediche; Azienda Ospedaliera Universitaria Senese – Ospedale Santa Maria alle Scotte Viale Mario Bracci, 53100 SIENA, ITALY**
- Contact email: **alessandra.renieri@unisi.it**

European Reference Network
for rare or low prevalence complex diseases
Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

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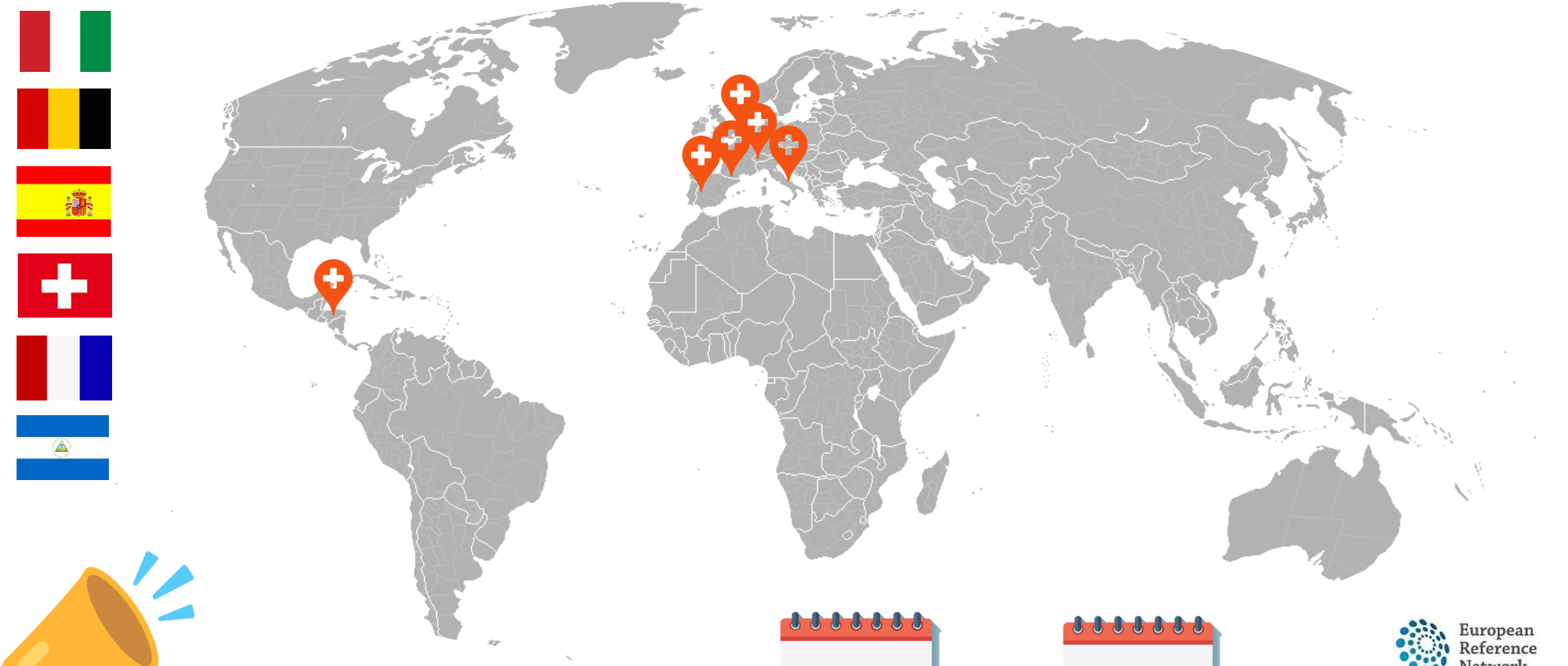

DATA COLLECTION from JUNE to SEPT.

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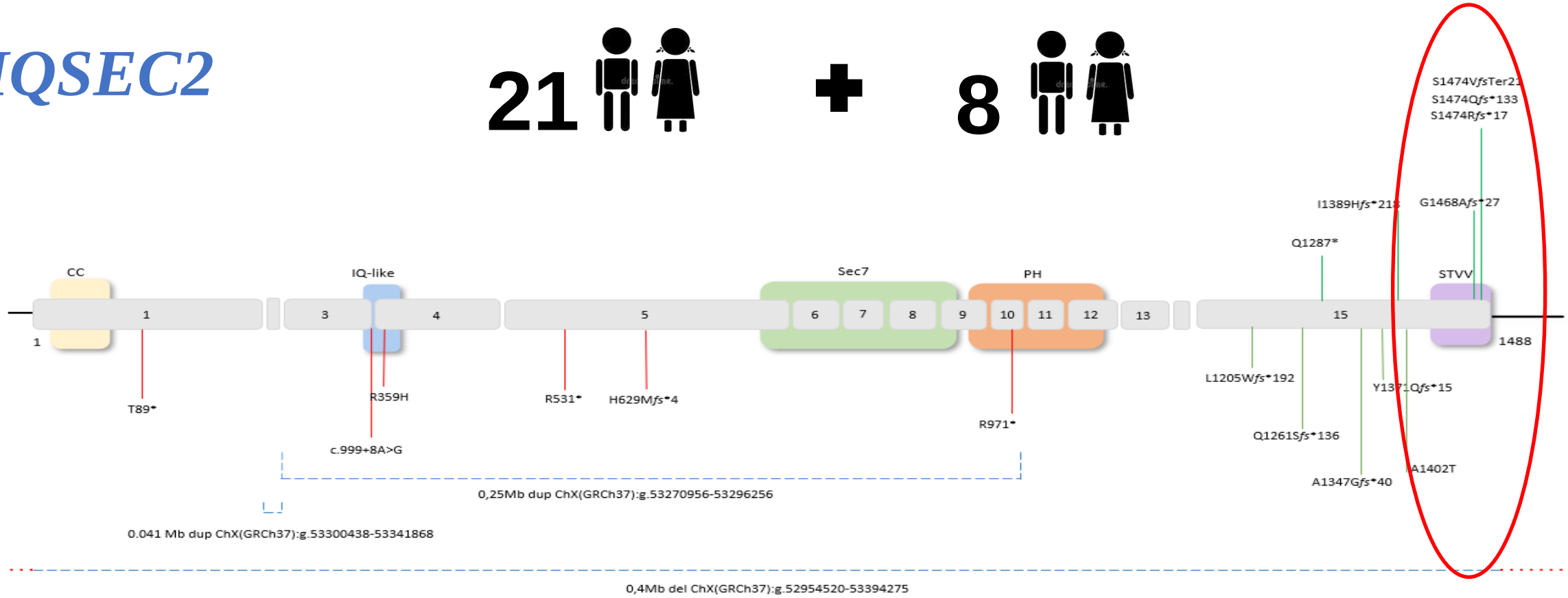
IQSEC2

21 + **8**



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Thanks for the collaboration!

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