European Reference Network

Endo-ERN

Transversal Working Group on Genetic Testing

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Transversal WG „Genetic Testing“

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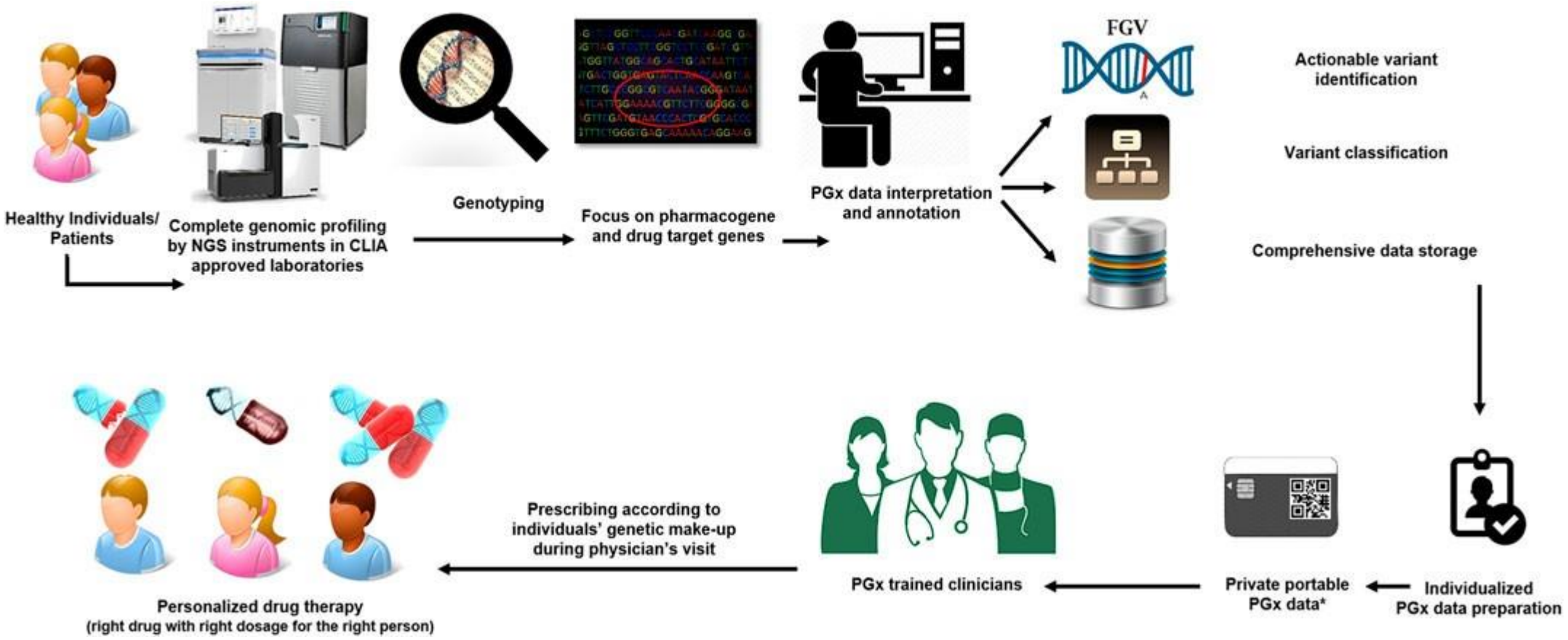
We have entered in the era of *omics* in research, diagnostics and therapy



Genetic testing at different ages: when and why?

- Prenatal
- Neonate (Newborn screening)
- Children
- Adults

www.nature.com/ejhg

ESHG

REVIEW ARTICLE

Check for updates

Rapid genomic testing for critically ill children: time to become standard of care?

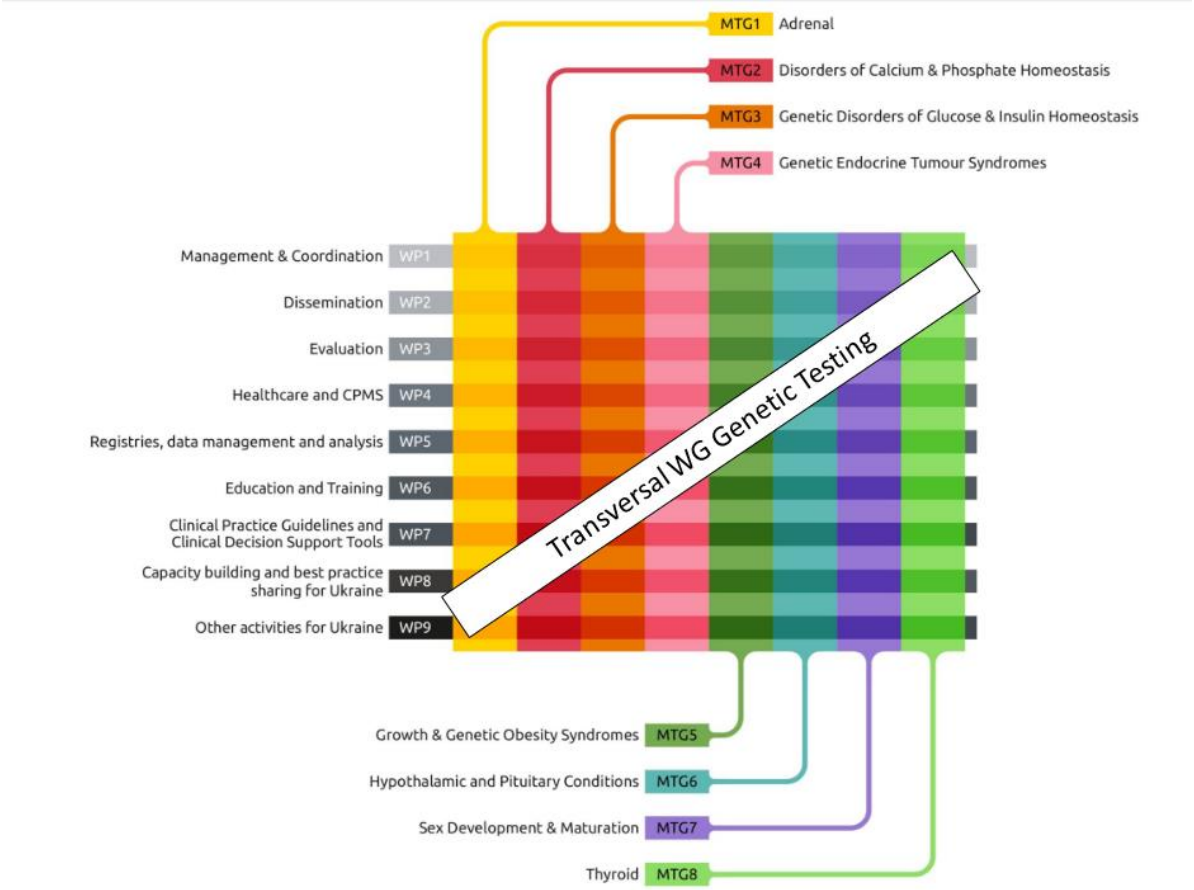
Zornitza Stark[1,2,3]✉ and Sian Ellard[4,5]

- *Pilot study for WGS in 37 patients with SRS features:*
  49% could be solved, including two adults
- *Example:* 34 y woman seeking advice due to progressing health issues in addition to SRS features. Diagnostic odyssee over more than 30 years.

"To aid in diagnosis of a chronic disease. A current medical condition may be caused by a previously unrecognized genetic defect, or the disease may have a genetic component."

Why do we need a transversal WG „Genetic Testing“ in our ENDO-ERN?

- The ENDO-ERN covers a broad range of endocrine disorders, and many entities are caused by genetic variants.
- These disorders are heterogeneous, but the genetic mechanisms behind them as well as the diagnostic testing strategies are very similar.



Why do we need a transversal WG „Genetic Testing“ in our ENDO-ERN?

- The need to implement and run diagnostic genetic tests in ENDO-ERN patients is obvious, and requires common activities in respect to offer these tests over Europe, to explain and use the results for the optimal benefit of our patients.
- Genetic testing has to be accompanied by quality assurance measures.

EMQN
The European Molecular Genetics Quality Network

Why do we need a transversal WG „Genetic Testing“ in our ENDO-ERN?

- The ENDO-ERN is continuously contacted by initiatives working in the field of genetics (SOLVE-RD, ESHG, …)

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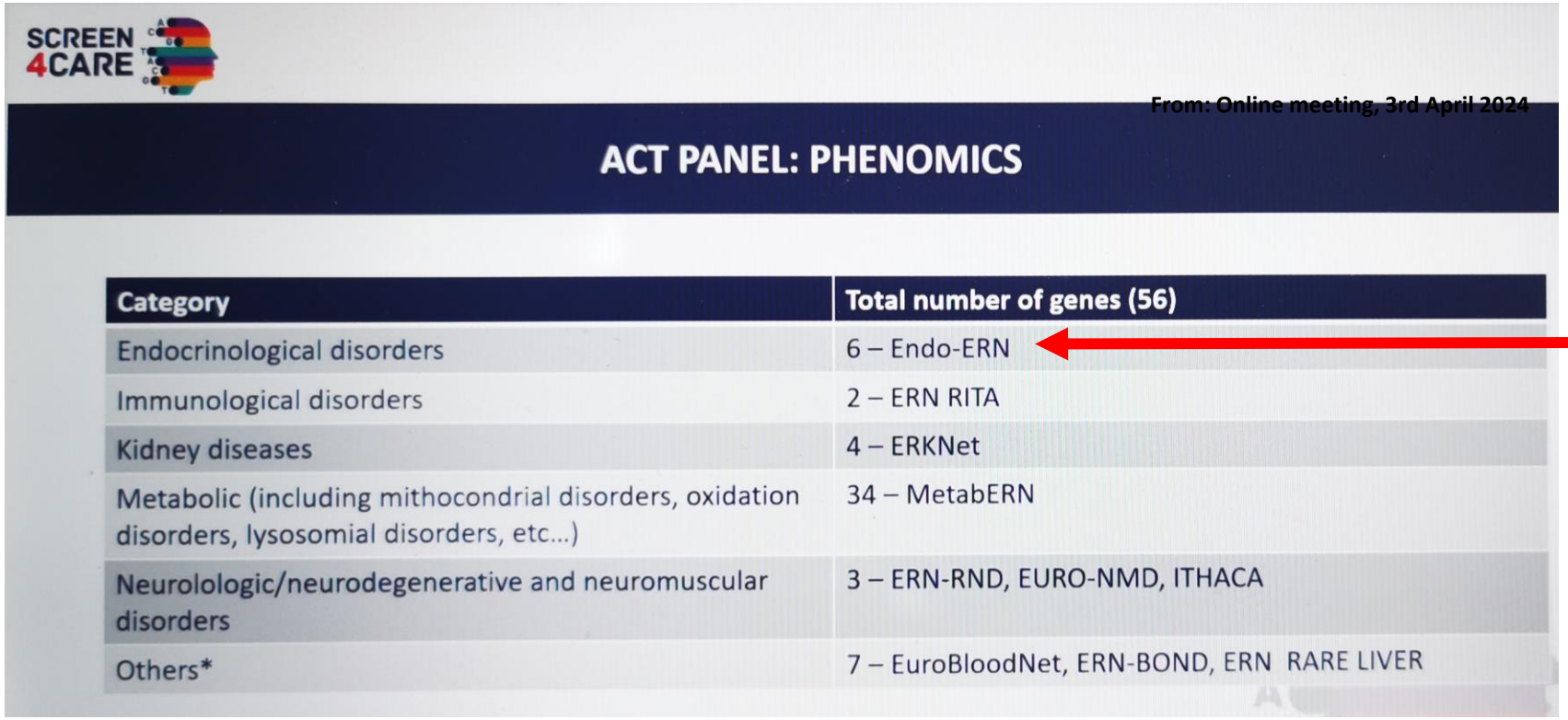

Selection of actionable genes could have been coordinated by transversal WG

Screen4Care’s is about to establish a rare disease focused genetic newborn screening, based on suggested genes from European experts and ERN members. This EU project receives support from the European Union’s Horizon 2020 research and innovation programme and EFPIA.

Goals and Outcomes

Goal	Outcome
Identify disease and/or genetic diagnostic expert in each MTG as contact person (invitation via the ENDO-ERN and actively contacting the MTG chairs)	Inclusion of all MTGs and all disorders
Identification of the needs of each MTG (Questionnaire)	Fill gaps in the available diagnostic testing, improve tests, increase the availability in different countries and health systems
Establish and harmonize monogenetic rare diseases and somatic testing strategies	Improve genetic testing (to detect e.g. mosaicism) Stimulate interdisciplinary discussions between different laboratory disciplines (i.e. geneticists, (molecular) pathologists)

Goals and Outcomes

Goal	Outcome
Evaluation of existing guidelines in the field of genetic testing	To improve guidelines, identify gaps and update where necessary
Development of guidelines for genetic testing in the field of endocrine disorders, either by participating in and contributing to already existing guidelines	Smooth and applicable genetic testing guidelines, which consider daily routine and the best standard for genetic testing. But: avoid double work and cooperate with other societies (e.g. ESHG)
Identify genetic methods and testing strategies which might belong to the basic diagnostic workup.	Suggest a standardized genetic testing regiment whenever possible to guarantee a standardized testing procedure all over Europe This also serves as the basis for curated patient registries
Interaction via CPMS/Training	Education/Training
Coordinated cooperation with players in the field of genetic testing, e.g. ESHG, Solve-RD (compare also goal “Guidelines”)	To orchestrate and to channel the activities of ENDO-ERN in the field of genetics, to avoid fragmentation and unwished developments in genetic testing (IVDR, commercial Direct-to-consumer genetic tests)

Timeline

- TE/DP presented their ideas to the ENDO-ERN steering committee on Feb 6th 2023

- A first invitation to all ENDO-ERN members to participate in the transversal WG had been sent out:
  Feedback from groups in BE, CZ, HU, IT, NE (clinicians, molecular diagnostics, pathology): "we think it is an excellent idea to create a transversal working group to address the aspects related to the genetics of rare endocrinological disorders. "

- Presentation of the transversal WG at the General Assembly in April 2024.

- In the course of/after the General Assembly, additional ENDO-ERN members can join.

- **Activity will start after GA by a virtual meeting of the colleagues interested in the WG, and a brain-storming. Via a questionnaire the need of our network in respect to genetic testing.**

Feel free to contact us at any time!

- Ideas/Suggestions/Contacts are welcome, preferably from every MTG

- **Contact:**

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