European Reference Network

Endo-ERN

Transversal WG „Genomic Testing“

GA 2026

Presenter name

Thomas Eggermann, Dirk Prawitt

Funded by the European Union

Status

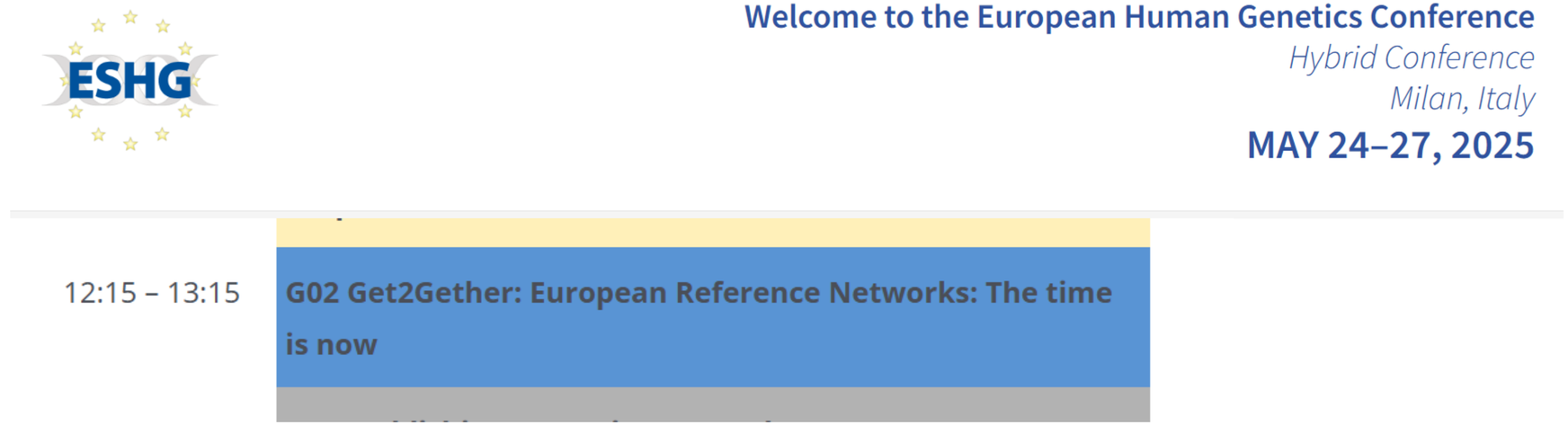
Currently 31 participants from 10 different countries

Virtual meetings: every 2-3 months, with the current focus on the joint paper

ERN-ESHG collaboration

ESHG – ERN collaboration

- communication managers (E. White) and liaison managers (TE, DP)
- Virtual meetings appr. every 3 months



- 2nd face2face meeting ESHG-ERN representatives planned at ESHG conference 2026

(ENDO) ERN - ESHG

European Reference Network

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Teaching & Masterclass

- ESHG becomes part of the ERNs course portfolios and vice versa
- Germline genetics needs to be part of the educational program of ERNs
- ESHG is willing to engage in education if the part of genetics is part of the ERNs educational programmes
- ERN may request collaboration from ESHG to support genetics-related educational contents (“not related to Industrial interests”).
- Developing ESHG-ERN Courses endorsed by ESHG and ERN.

Cross-referencing in ESHG and ERNs websites regarding collaborative actions, joint-educational initiates and liaison members.

Visibility & Communication

•**Visibility : ERN visible on the ESHG website and vice versa**

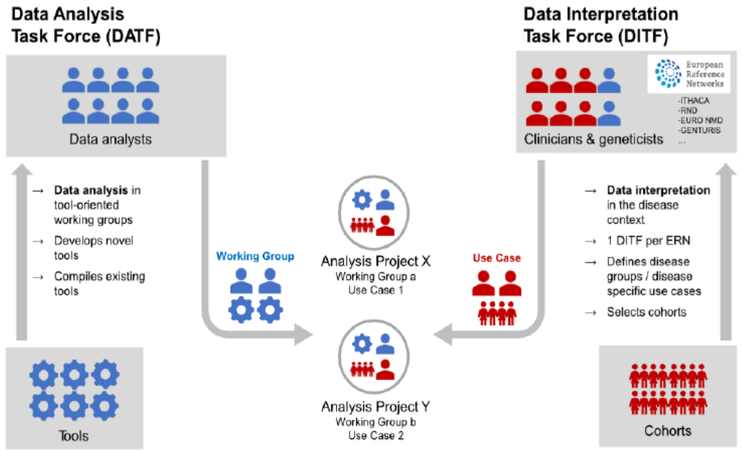
How to make ERNs visible on the ESHG website?
How to make ESHG visible on the ERN Websites?

Visibility of ERN-ESHG **liaison members on both websites?**

Communication managers (for ERN-ESHG information flux) and inform the ESHG Office (Jerome del Pichia) ?

Update ERDERA
Diagnostic Research Workstream

Joint expert re-analysis



WHAT CAN YOU DO AS ERN TO CONTRIBUTE TO THE DRW?

- Inform your HCPs to contribute samples to the DRW
- Have HCPs sign up to contribute to the ERN-specific DITF if they have expert knowledge on:
  - ✓ Data interpretation for (non-)coding variants
  - ✓ Specific genes or phenotypes of relevance to your ERN
  - ✓ Novel gene/mechanism/paradigm discovery
  - ✓ (Data analysis to join DATF working groups)
- Nominate a candidate for the position ERN-specific DITF lead and provide the information to Holm Graessner and Lisenka Vissers

The WG Genetic Testing aims to moderate the cooperation between the HCPs of our network and the ERDERA Diagnostic workstream group, e.g. by bringing them in contact with ERDERA, and by exchanging information and experiences on national level in respect to the legal issues (e.g. data protection).

In addition to these individual cooperations between our HCPs and ERDERA Diagnostic Workstream, our ENDO-ERN has to assign colleagues for the Data Interpretation Task Force (DIFT).

We have been advised to encourage the MTG chairs to promote this in the network. If all agree, the follow up dissemination to HCPs and identification of Endo-ERN representation for the Data Interpretation Task Force, can be done afterwards by our coordination office.

Is it possible to ask each MTG to nominate at least one representative to discuss further steps with the WG (deadline: Feb. 15th 2026: no answers)

ERDERA2026 call

Conceptual phase of Calls for Proposals 2026

“Resolving unsolved cases in rare genetic and non-genetic diseases”

Submission deadline for pre-proposals: February 12th, 2026

Currently pre-proposals submitted

CHH (Marco Bonomi)

RTH (Luca Persani)

(DSD (Martin Kircher))

Checklist for Ethical, Clinical, and Operational Evaluation of Genetic Testing Offers

Initiative by Natasha Appelman-Dijkstra, Corinna Grasemann, Eva Kassi, Martha Kirchhoff

Purpose

This document provides a structured checklist to support clinicians, genetic laboratories, ERN working groups, and affiliated stakeholders in evaluating genetic testing offers. It aims to ensure that testing initiatives meet appropriate standards for scientific rigor, patient safety, ethical integrity, and compliance with European regulatory frameworks.

Scope

The checklist addresses scientific validity, laboratory standards, data protection, ethical considerations, clinical responsibilities, financial transparency, and institutional oversight. It can be used during project planning, review of industry collaborations, or preparation for clinical implementation.

Checklist for Ethical, Clinical, and Operational Evaluation of Genetic Testing Offers

Status:

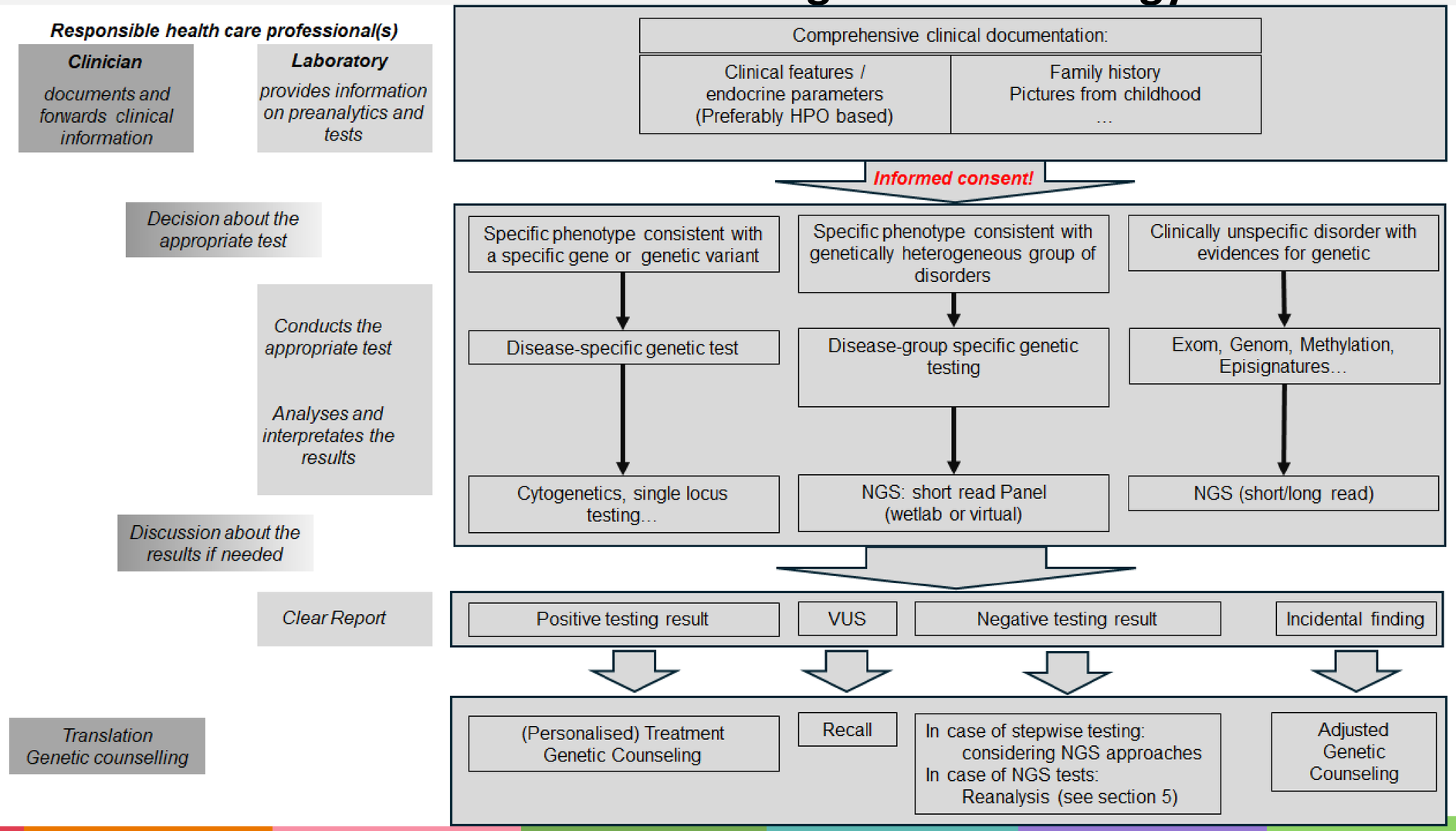
MTG 2 has prepared hypoparathyroidism specific items

Place them on the MTG 2 website (if chairs Kassi and Grasemann agree)

Maybe link it to the hypoparathyroidism module in EuRREB

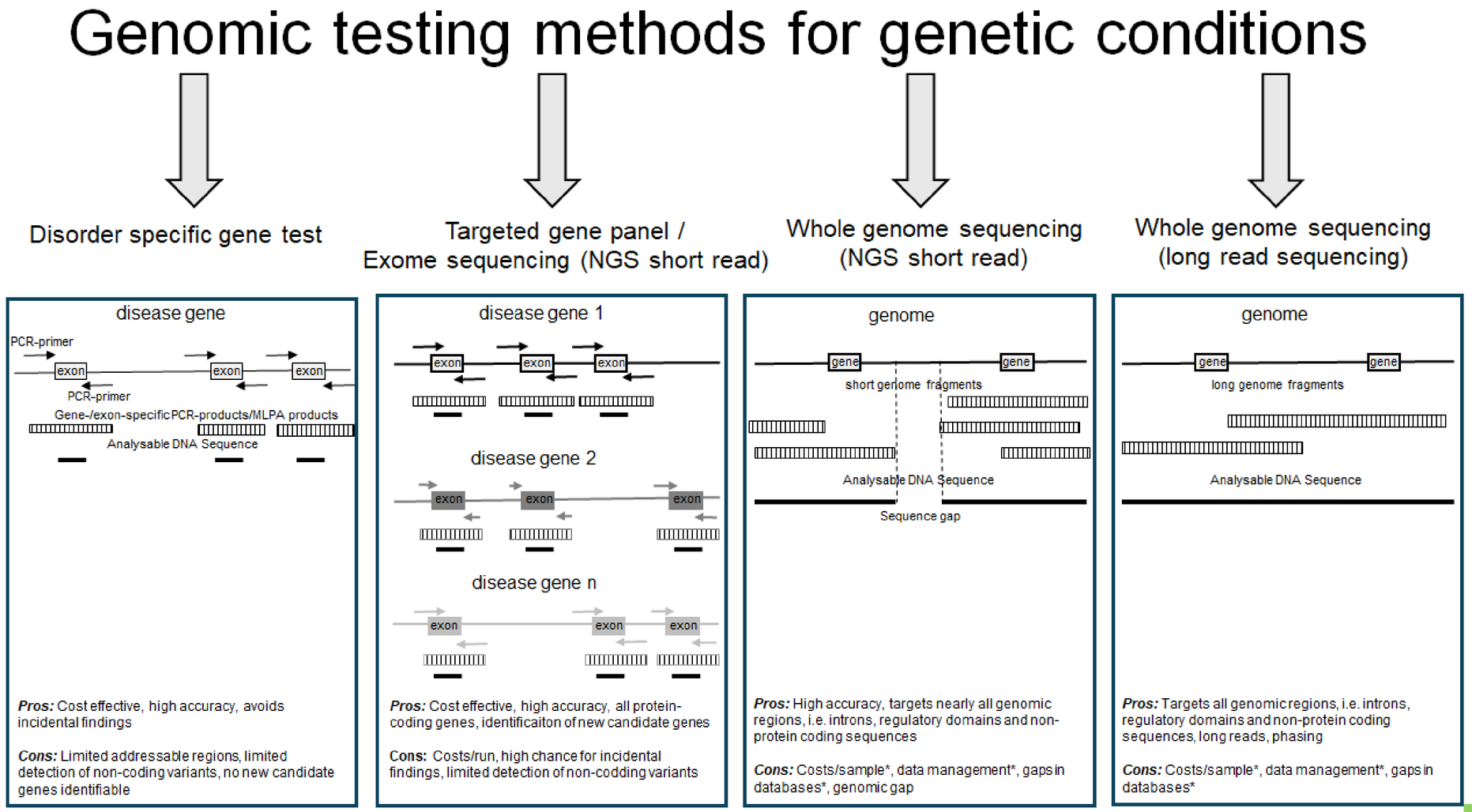
Joint paper (close to submission):
„ Genetic testing and reporting: What the endocrinologists should know and can expect”

Decision tree for molecular testing in endocrinology



Joint paper (close to submission):
„ Genetic testing and reporting: What the endocrinologists should know and can expect”

NGS strategies and their major pros and cons.



Joint paper (close to submission):
„ Genetic testing and reporting: What the endocrinologists should know and can expect”

Example for the design of a structured report

Lab-Identifier, Date of Report

Genetic Report

Name, Prename:	**Doe, Jane**	**Report-No.:**	**DOE-ID-RE**
Date of birth	**01.01.2022**	**DNA-No.:**	**26-D01**
Sex:	Female	Request received:	16.01.2026
Sample type:	EDTA-Blood	Your ID:	

Reason for referral:

Genetic testing for short stature. The patient was born SGA, and shows proportionate growth retardation (<P3), a frontal bossing, a pointed chin and clinodactyly V. Preceding testing for Silver-Russell syndrome was negative (see our report 26-MLPA-1). The mother shows similar clinical features.

Test methodology: **Exome analysis**

Result: **Conspicious finding**

Jane Doe is heterozygous for the pathogenic sequence variant in the *HMGA2* gene, NM_003483.6(HMGA2):c.193C>T (p.Gln65Ter) (see variant detail).

Interpretation:

The genetic finding of the pathogenic *HMGA2* variant is compatible with the clinical features of Jane Doe.

Pathogenic variants in *HMGA2* are associated with growth retardation phenotypes (OMIM#618908) and are inherited in an autosomal dominant (reference 1). Accordingly, the patient will inherit the variant with a chance of 50%. As mentioned before, the mother shows similar clinical features, therefore testing of a maternal sample should be considered. In case the mother is carrier of the *HMGA2* variant, the chance for further children of the mother of your patient is 50% as well.
For clinical management we refer to reference 2.

Genetic counselling is suggested.

Signature of the responsible laboratory staff members

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Variant details:

Gene	Variant description (HGVS)	zygosity	Variant type	Allele frequency (gnomAD)	Bioinformatic predition	ClinVar ID	Classificatio (ACMG)
HMGA	NM_003483.6: c.193C>T p.Gln65Ter Chr12 (hg38): g.49022066G>A (hg38)	heterozygous	Nonsense	0.05%	CADDphred: 35.9 REVEL: 0.067	253034	**Pathogenic** (PVS1, PM2_supporti PP3_moderat

Comment on the variant:
Information from other databases and literature. Reason for classification.
This variant has not yet been reported in the literature or in the ClinVar database. It has not yet been detected in the general population (gnomAD v4.1.0) (PM2_sup). Bioinformatic prediction programs classify the variant as potentially disease-causing (PP3_mod).

References:
Methods and Limitations:

- Page 2 -

Example for a virtual precise and comprehensive genetic report.

Note that the information relevant for the referring clinician is summarized on the first page,

technical information about the variant and the methods are listed on the following page.

Joint paper (close to submission):
„ Genetic testing and reporting: What the endocrinologists should know and can expect”

General overview on the aspects which should be considered for reporting

Sample identification:

- Type of incoming sample
- Each sample from the same patient must be identified separately for traceability

Reason for request:

- Clinical diagnosis / Disease name: clinical symptoms as comprehensive and as precise as possible, preferably allowing the transformation into HPO
- Reason for referral (diagnostic, prenatal testing, carrier testing etc.)
- Reference to previous reports if available
- Reference to family history

Specification of the applied test (might be provided at the end of the report)

- Precise but short information on the diagnostic assay(s), i.e. manufacturer or in-house test, kit version, platform, bioinformatic pipeline, analysis software
- cope of the assay(s), e.g.:
  - Targeted genomic region (reference genome and/or transcript (preferably MANE, in case of NGS: read depth)
  - Type of addressed genetic variants (e.g. SNVs, CNVs/SVs, Repeats)
  - Sensitivity and precision
  - If specific genes have been requested, the laboratory should explicitly address these in the report; any analytical gaps in these genes should be named.
  - Additional limitations of the assay
  - The report must clearly state which variants are not reported (e.g: Benign variants, likely benign variants, VUS).

Results:

- Variant nomenclature must be based on the HGVS and – in case of CNVs – on the ISCN recommendations
- Variant classification should be based on ACMG criteria
- List of databases and literature references to further explain the clinical relevance of (probable) pathogenic variants.
- Unreported benign variants, likely benign variants, and additional VUS should be available upon provider or patient request.
- Variants in genes that are not known to be associated with a phenotype (genes of unknown significance, GUS) should not be mentioned.
- If the laboratory chooses to report a variant in a GUS (e.g., a de novo variant in a constrained gene without a reported disease association), then the lack of disease association should be noted and it should be reported as a VUS in a section separate from the primary findings.

Interpretation:

- Clear and concise statement about the relevance of the variants listed, taking into account the reason for request.
- Consideration should be given to whether further investigations could provide more information about the potential medical relevance of the VUS.
- In case an incidental/secondary finding has been identified, this should be mentioned, but detailed information should be provided in a separate report.
- Optional: Information about available guidelines and recommendations for clinical management.
- If necessary, the report might mention the option of a possible reclassification of VUS after a specified period, e. g. in case of newly presenting symptoms (see section 5).
- In case the reporting lab is a aware of further genetic tests, e.g. in the prenatal period, this should be mentioned to enable the referring clinician to join the information.

Joint paper (close to submission):
„ Genetic testing and reporting: What the endocrinologists should know and can expect”

Status:

- **Final version is currently circulating in the WG team (deadline May 15th)**
- **Journal in discussion with the SCAB**

Contribution to the

European Reference Network

Endo-ERN

European Society of Endocrinology

ECE 2026

9-12 MAY 2026

Prague, Czech Republic

28th European Congress of Endocrinology

11:20 - 12:50 CEST

Endo-ERN Session

Symposium

Panorama Hall

Introduction on Endo-ERN initiatives (with a focus on the EndoCompass paper)

Tuesday, 12 May
11:20 - 11:20 CEST

Add to my programme | Save calendar file (.ics)

Panorama Hall

Transversal working group on Genomic Testing

Thomas Eggermann (Aachen, Germany)

Tuesday, 12 May
11:20 - 11:40 CEST

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Panorama Hall

The peadiatric thyroid cancer registry

Hanneke van Santen (Utrecht, The Netherlands)

Tuesday, 12 May
11:40 - 12:00 CEST

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Panorama Hall

The reduced life expectancy and treatment options for RTHbeta

Irene Campi (Milano, Italy)

Tuesday, 12 May
12:00 - 12:20 CEST

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Panorama Hall

The Psychosocial consequences of living with a pituitary condition.

Johan De Graaf (Nijkerk, The Netherlands)

Tuesday, 12 May
12:20 - 12:40 CEST

Add to my programme | Save calendar file (.ics)

Webinar

REGISTER: Endo-ERN X Endocrine Connections Genetic Testing Approaches as the Basis for Personalized Medicine

This webinar will be on June 10th, 2026

Save the date and register now for the Endo-ERN X Endocrine Connections ***Genetic Testing Approaches as the Basis for Personalized Medicine*** taking place Wednesday 10 June at 16:30-17:30.

The Changing Landscape of Genetic Testing in Rare Endocrine Conditions (15 min)

Prawitt/Eggermann

From Testing to Tailored Management (15 min)

Van der Kaay

European Collaboration and Future Perspectives (15 min)

Persani

Feel free to contact us at any time!

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

Contact:

Thomas Eggermann
(teggermann@ukaachen.de)

Dirk Prawitt
(dprawitt@uni-mainz.de)