Checklist for Genetic Testing Eligibility in Idiopathic Hypoparathyroidism

Guidance Note for Healthcare Providers (HCPs)

Use of the Checklist for Genetic Testing Eligibility in Idiopathic Hypoparathyroidism

This checklist is designed to support healthcare providers in the **systematic collection of clinical, biochemical, and family data** for patients with suspected idiopathic hypoparathyroidism who may be candidates for genetic testing.

Purpose: The aim is to ensure that all relevant information is available prior to **CPMS case discussion**, facilitating:

- Accurate eligibility assessment for genetic testing
- Efficient multidisciplinary review
- Harmonized decision-making across the ENDO-ERN network

How to Use This Checklist

I. Complete all sections as thoroughly as possible before CPMS submission.
II. Provide actual laboratory values with reference ranges where applicable.
III. Tick all relevant boxes and add additional details where needed.
IV. If information is unavailable, indicate this clearly (e.g., “not performed” or “unknown”).

Key Requirements

I. Secondary causes must be excluded before classifying a case as idiopathic.
II. Pay particular attention to identifying ***syndromic features*** and ***family history,*** as these are critical for genetic evaluation.
III. Ensure that the dataset is complete prior to CPMS discussion, as incomplete data may delay decision-making.

After Completion

I. Share the completed checklist during **CPMS case presentation**.
II. If genetic testing is recommended, proceed with:
   a. Patient information and consent
   b. Sample collection and logistics
III. Ensure all data and outcomes are documented in CPMS and relevant registries.

Important Note

Genetic testing should only be initiated following **CPMS consensus recommendation**. This checklist is a key tool to support that decision-making process.

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Patient Identification & Demographics

Patient ID / initials: _____
Date of birth / age: _____
Sex: _____
Ethnicity: _____
Referring center / clinician: _____

2. Clinical Presentation (Phenotype)

- Age at onset of hypocalcemia / hypoparathyroidism:
- Symptoms at presentation (tick all that apply):

☐ Seizures
☐ Tetany
☐ Paresthesias
☐ Cardiac arrhythmia
☐ Asymptomatic (incidental)

3. Biochemical Profile

- Serum calcium: _____
- Serum phosphate: _____
- PTH: _____
- Magnesium: _____
- Vitamin D (25-OH): _____
- Renal function: _____
- 24h Urinary calcium: _____

4. Exclusion of Secondary Causes

(MANDATORY before labeling "idiopathic")

☐ Post-surgical hypoparathyroidism excluded
☐ Autoimmune causes evaluated
☐ Magnesium deficiency excluded
☐ Vitamin D deficiency excluded
☐ Infiltrative diseases excluded
☐ Drug-induced causes excluded

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5. Syndromic / Extra-Parathyroid Features

(Important for identifying genetic/syndromic forms, tick all that apply)

☐ Dysmorphic features
☐ Developmental delay / intellectual disability
☐ Cardiac anomalies
☐ Renal anomalies
☐ Hearing loss
☐ Skeletal abnormalities
☐ Immune dysfunction / recurrent infections
☐ Other endocrine disorders.....

6. Imaging & Additional Investigations

☐ Renal ultrasound (nephrocalcinosis / stones)
☐ Brain imaging (basal ganglia calcifications)
☐ Cardiac evaluation (if indicated)
☐ Bone density (DXA, if available)

7. Family History

Consanguinity: _____
Affected relatives: _____
Pedigree available: _____

8. Previous Genetic Testing

☐ None
☐ Gene panel (specify):
☐ Whole exome/genome sequencing
- Results (if available):

☐ Pathogenic / likely pathogenic
☐ Variant of uncertain significance (VUS)
☐ Negative

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9. Treatment & Disease Behavior

Tick all that apply:

Current treatment:

- ☐Calcium supplementation
- ☐Activated vitamin D analogues
- ☐PTH analogues

Treatment response:

- ☐ Stable
- ☐ Difficult to control
- ☐ Refractory

Complications:

- ☐Hypercalciuria
- ☐Nephrocalcinosis
- ☐Renal impairment

10. Eligibility Criteria for Genetic Testing

(To be confirmed during CPMS discussion) Tick all that apply:

- ☐ Idiopathic hypoparathyroidism confirmed
- ☐ Early-onset disease
- ☐ Syndromic / atypical features
- ☐ Positive family history
- ☐ Unusual or refractory disease course

11. Administrative & Process Requirements

- ☐ Case discussed with MTG2 Chairs prior to CPMS submission
- ☐ Case entered into CPMS with full dataset
- ☐ Contact with genetic testing working group *(email: myavropoulou@med.uoa.gr)*

12. Consent & Logistics (Post-Eligibility)

- ☐ Patient informed about genetic testing
- ☐ Written informed consent obtained
- ☐ Sample collection arranged
- ☐ Shipment logistics confirmed

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