



1st Workshop

Codification of suspected/undiagnosed rare diseases

WP 5-Team

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★ <http://rd-code.eu>

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Coding of undiagnosed rare diseases patients in HER: existing experiences and future perspectives

- > Existing experience of coding situation
- > Of undiagnosed or suspected rare diseases patients
- > Guidelines to be proposed
- > Collaboration with SOLVE-RD

Final goal (Sept 2020?): consensual documentation on codification of suspected/undiagnosed rare diseases

What has been done so far

- > **Done** **Milestone 5.3:** *Collection of existing experiences about the coding of undiagnosed or suspected rare diseases patients in Electronic Health Records*
 - ★ Sent to all collaborating partners to collect experiences
 - ★ Sent to implementing countries
 - ★ Sent to SOLVE-RD
- > Difficulty to understand the document scope: it is not about research initiatives, but about how to identify undiagnosed patients in an EHR /database
- > Few feedbacks as the subject is quite advanced
- > **Delayed** **Milestone 5.4:** final draft (initial due date: May 20)

JRC set of common data elements of RD registration

GROUP	ELEMENT N°	ELEMENT NAME	ELEMENT DESCRIPTION	CODING	COMMENT
6 Diagnosis	6.1.	Diagnosis of the rare disease	Diagnosis retained by the specialised centre	Orpha code (strongly recommended – see link) / Alpha code/ ICD-9 code/ ICD-9-CM code / ICD-10 code	http://www.orphadata.org/cgi-bin/inc/product1.inc.php
	6.2.	Genetic diagnosis	Genetic diagnosis retained by the specialised centre	International classification of mutations (HGVS) (strongly recommended – see link) / HGNC / OMIM code	http://www.hgvs.org
	6.3	Undiagnosed case	How the undiagnosed case is defined	<ul style="list-style-type: none">• Phenotype (HPO)• Genotype (HGVS)	



> Broad recommendations:

★ Description of phenotype and/or genotype

> Can be interpreted and implemented in different ways

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Recommendation #1

HOW TO INDICATE THAT THE PATIENT IS UNDIAGNOSED?

Identification of an undiagnosed patient (1)

Recommendation #1

Standard procedure and guide for coding with Orphacodes, Guideline 3 (RD-ACTION):

Whenever possible capture the information of the diagnostic assertion for all RD cases. Use the Options “Suspected rare disease”, “Confirmed rare disease” and “Undetermined diagnosis”. Additional options might be helpful.

=> This recommendation is valid to identify the undiagnosed patients in ERH

Identification of an undiagnosed patient (2)

> Is there a way to identify undiagnosed patients in HIS, **without adding a new item?**

TO BE DISCUSSED:

- ★ Can we use high level of Orphacodes (group of disorders)? And which level?
- ★ Should a specific Orphacode be created?

Orphanet classification of rare renal diseases

> [Rare renal disease](#) ORPHA:93626

└ [Glomerular disease](#) ORPHA:93548 -

└ [Collagen-related glomerular basement membrane disease](#) ORPHA:544590 -

└ [Alport syndrome](#) ORPHA:63 +

└ [HANAC syndrome](#) ORPHA:73229

Groups of disorders

Disorders

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Recommendations #2 & #3

HOW TO DESCRIBE UNDIAGNOSED PATIENTS?

Description of an undiagnosed patient

Phenotype: HPO + Genotype: HGVS

Recommandation #2

Each undiagnosed patient should be described by its phenotype, using HPO. When available, the genotype should be associated to help future diagnosis.

Additional phenotypic descriptors could be used (for instance Orpha groups of diseases; ICD; SNOMED...) as well as genetics descriptors (HGNC genes, OMIM...).

Promoting the use of Orphacodes

- Orphacodes should not be associated to the diagnosis level of assertion:
 - ★ We do not recommend to create codes such as:
 - > Genetically undetermined congenital cataract
 - > Intellectual disability of unknown cause
- However, when combined with the diagnostic assertion, coding with the broader Orphacodes (groups or categories) can be part of the phenotypic description

Orphacodes as a phenotypic descriptor

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Groups of disorders

Disorders

ORPHA:544590 [Group of disorders](#) [Collagen-related glomerular basement membrane disease](#)

[More information](#)

Keyword(s): Glomerular basement membrane disease due to a COL4A mutation

ORPHA:183586 [Group of disorders](#) [Genetic glomerular disease](#)

[More information](#)

ORPHA:93548 [Group of disorders](#) [Glomerular disease](#)

[More information](#)

Description of an undiagnosed patient - Orphacodes

Recommendation #3

- *The level of the Orpha classification (i.e. Group of disorders; disorder; subtype) should be used in the Orphanet datasource for all entries in association with the Orphacodes.*

Distinction between uncodable diseases and undiagnosed patients

> Uncodable diseases :

- ★ new or ultra-rare diseases
- ★ Not yet available in the Orphanet nomenclature

How to ask for a new ORPHAcode entry

> Go to the GitHub:

<https://github.com/orphanet-rare-diseases-issues/RD-CODE>

Do you agree on those recommendations?

Recommendation #1 :

Whenever possible capture the information of the diagnostic assertion for all RD cases. Use the Options “Suspected rare disease”, “Confirmed rare disease” and “Undetermined diagnosis”. Additional options might be helpful.

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Groups of disorders

Disorders

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