

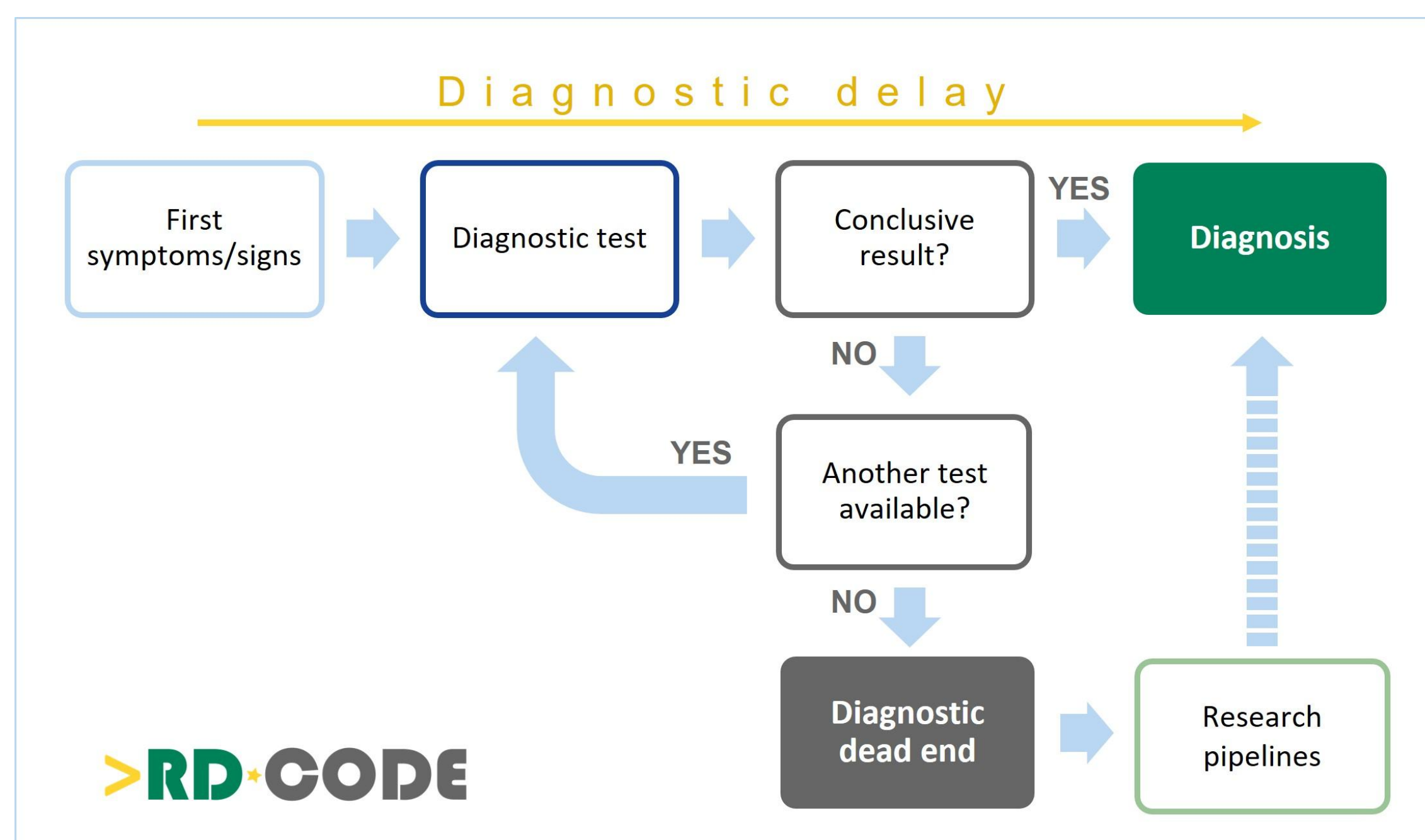
Undiagnosed rare disease patients: A dedicated ORPHAcode to make them count

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Introduction

★ As expertise is scarce and rare diseases (RD) are complex, RD patients can remain undiagnosed for many years. The period of searching for a diagnosis, called **diagnostic delay**, sometimes leads to a **diagnostic dead end** when the patient's disease is impossible to diagnose after undergoing all available investigations.



★ In recent years, extensive efforts have been made to support the implementation of ORPHA nomenclature in health information systems (HIS) such as electronic health records and/or registries so as to allow RD coding. Until recently, the nomenclature only encompassed codes for specific RD. Persons suffering from a suspected RD who could not be diagnosed even after full investigation, could not be coded with ORPHAcodes.

Methodology

- ★ The RD-CODE project aimed at making those RD undiagnosed patients identifiable in HIS to produce crucial epidemiological data.
- ★ The RD-CODE multi-stakeholder panel of experts produced three recommendations for the coding of undiagnosed RD patients :
 - > Capture the diagnostic ascertainment for all RD cases
 - > Use the newly created ORPHAcode **ORPHA:616874** (see below)
 - > Use additional descriptors in registries

Undiagnosed RD patients were defined as patients for whom :

- > no clinically-known disorder could be confirmed by an expert center
- > after all reasonable efforts to obtain a diagnosis
- > according to the state-of-the-art and diagnostic capabilities available

Clinical diagnosis	Etiological diagnosis	Diagnostic status for RD-CODE
Established	Established	Diagnosed
Established	Unknown	Diagnosed
Suspected	Established	Undiagnosed
Suspected	Unknown	Undiagnosed
Unknown	Established	Undiagnosed
Unknown	Unknown	Undiagnosed

ORPHA:616874

“Rare disorder without a determined diagnosis after full investigation”

Available in the Orphanet nomenclature, together with the guidelines to ensure its correct and homogeneous use.

This new ORPHAcode could be a **game-changer** for patients, clinicians and researchers in the field, enabling:

- > assessment of the RD population, including undiagnosed patients,
- > adaptation of policy measures including financing for care and research programs,
- > improved access of undiagnosed patients to research programs.

Rare disorder without a determined diagnosis after full investigation Suggest an update

Disease definition

A rare disorder for which all reasonable efforts have been done by rare diseases experts to determine a diagnosis according to the state of the art and available diagnostic capabilities, but did not enable to conclude on a clinically known concept. It is recommended to restrict the use of this entity for coding purposes to rare disease experts.

A code for specific and unambiguous designation of undiagnosed patients

ORPHA:616874

Classification level: Disorder

Synonym(s): Fully investigated rare disorder without a determined diagnosis

Prevalence: -

Inheritance: -

Age of onset: -

ICD-10: R69 UMLS: C5680389

ICD-11: MG48

A summary on this disease is available in [Français](#) (2021) [Español](#) (2021) [Deutsch](#) (2021) [Italiano](#) (2021) [Português](#) (2021) [Nederlands](#) (2021)

Guidelines to properly use this new code

- > The code must **NOT** be used for diseases that are not (yet) available in the Orphanet classification
 - ★ If a code is not available, please use Github, the online ticketing system that allows requests to be stored, tracked and made available to others. (<https://github.com/orphanet-rare-diseases-issues/RD-CODE>).
- > The code must **NOT** be used for coding patients along their diagnostic pathway
 - ★ Make sure to use this code only after all reasonable efforts to obtain a diagnosis according to the state-of-the-art and diagnostic capabilities available were done.
- > Because of the latest point, it is recommended that **only experts** (i.e. RD centres where available) **should be allowed to assign this code**
- > The use of this code can be combined with other descriptors

Guidelines video ->

<http://www.rd-code.eu/guidelines-for-coding-undiagnosed-patients-video/>



Socially and psychologically empowering

Grant access to scientific advances

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