



# RD-CODE Workshop Country Presentation

## Spain

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on behalf of the Spanish collaborating stakeholders

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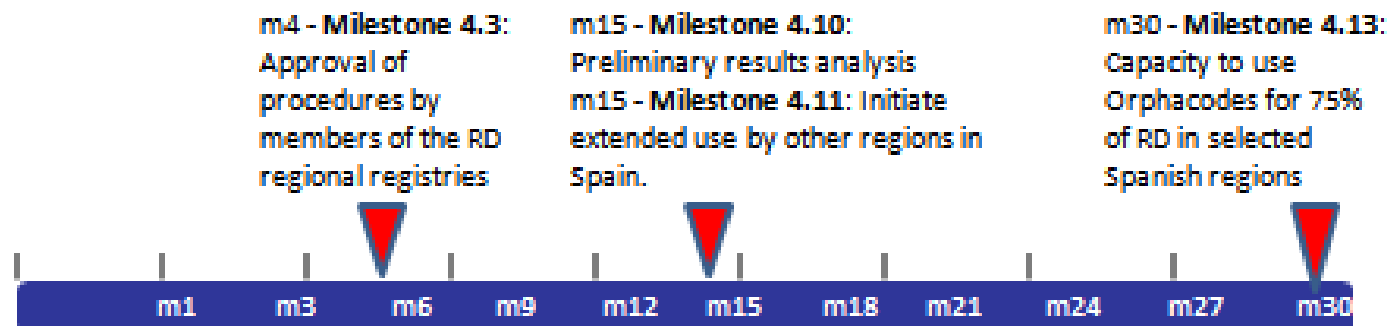
★ <http://rd-code.eu>



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# Timeline



## Phase 1

**Pilot phase:** Use of the “Standard procedure and guide for the coding with Orphacodes”  
And the “Specification and implementation manual of the Master file” by a selected group of regions (6)

## Phase 2

► m15 – Deliverable 4.4: Preliminary results analysis

## Phase 3

**Extended** use of the “Standard procedure and guide” and “Specification and implementation manual of the Master file” by other regions/national level based on the results obtained in Phase 1 (**at least 2**)

## Phase 4

► m30 – Deliverable 4.11: Final Report  
including **Recommendations** for implementation  
at a national level

The regional registries setup

# THE STARTING SITUATION

# Regional Rare Diseases registries

- > Catalonia (7.5 M)
- > Basque country (2.2 M)
- > Navarre (640 K)
- > Castile and Leon (2.5 M)
- > Valencian Region (5 M)
- > Murcia (1.5 M)

*Near 19 million people*

- ✓ Minimum Basic Data Set
- ✓ Mortality
- ✓ Morbidity
- ✓ Congenital anomalies
- ✓ Renal diseases
- ✓ Genetics
- ✓ Primary care
- ✓ Physicians
- ✓ Medical records
- ✓ Drugs registry
- ✓ Others

# Regional Rare Diseases registries

- > Catalonia (7.5 M)
- > Basque country (2.2 M)
- > Navarre (640 K)
- > Castile and Leon (2.5 M)
- > Valencian region (5 M)
- > Murcia (1.5 M)

## ✓ Population-based registries

- ✓ ICD-9-CM
- ✓ ICD-10
- ✓ ICD-10-ES
- ✓ ICD-10-BPA
- ✓ ERA-EDTA
- ✓ Snomed-CT
- ✓ ORPHAcodes

# Regional Rare Diseases registries

	ICD-9-CM	ICD-10	ICD-10-ES	ICD-10-BPA	ERA-EDTA	Physicians
<b>Basque Country</b>			X			X
<b>Catalonia</b>	X	X	X		X	X
<b>Castile and Leon</b>	X		X		X	
<b>Murcia</b>	X		X		X	
<b>Navarre</b>	X	X	X			X
<b>Valencian Region</b>	X	X	X	X	X	

The process of establishing equivalences

# ICD-10-ES CODES PROPOSAL

# Mapping ORPHAcodes to ICD-10-ES

OrphaNumber	Name	Synonym	ICD-10 V2016
36	Acrocallosal syndrome		Q04.0
36	Acrocallosal syndrome	ACS	
37	Acrodermatitis enteropathica		E83.2
37	Acrodermatitis enteropathica	Acrodermatitis enteropathica, zinc deficiency type	
37	Acrodermatitis enteropathica	AEZ	
37	Acrodermatitis enteropathica	Inherited zinc deficiency	
38	Acrokeratoelastoidosis of Costa		Q82.8
38	Acrokeratoelastoidosis of Costa	AKE	
38	Acrokeratoelastoidosis of Costa	PPKP3	
38	Acrokeratoelastoidosis of Costa	Punctate palmoplantar hyperkeratosis type 3	
38	Acrokeratoelastoidosis of Costa	Punctate palmoplantar keratoderma type 3	
39	Acromelanosis		L81.4
40	Acromesomelic dysplasia, Maroteaux type		Q77.8
41	Dyschromatosis symmetrica hereditaria		L81.8



Lista tabular

## eCIE10ES

Edición electrónica de la CIE-10-ES Diagnósticos

2ª Edición-Enero 2018

Versión electrónica actualizada con corrección de erratas a diciembre de 2018

CLASIFICACIÓN INTERNACIONAL DE ENFERMEDADES  
10.ª REVISIÓN, MODIFICACIÓN CLÍNICA, EDICIÓN ESPAÑOLA  
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Dirección General de Salud Pública, Calidad e Innovación  
Subdirección General de Información Sanitaria  
[www.mscbs.gob.es](http://www.mscbs.gob.es)

PUEDE CONSULTAR CIE-10-ES 3ª EDICIÓN 2020 DESDE EL SIGUIENTE [enlace](http://rd-code.eu)

Para la notificación de incidencias técnicas relacionadas con un funcionamiento incorrecto de consulta eCiemaps, puede dirigirse al Servicio de Atención al Usuario a través de la siguiente dirección de correo electrónico: [cau-servicios@mscbs.es](mailto:cau-servicios@mscbs.es). Debe indicar sus datos de contacto (teléfono y mail) y el problema que se ha detectado.

orphanet

WE ARE THE 300 MILLION

#RareDiseaseDay

20 FEBRUARY 2020

The portal for rare diseases and orphan drugs

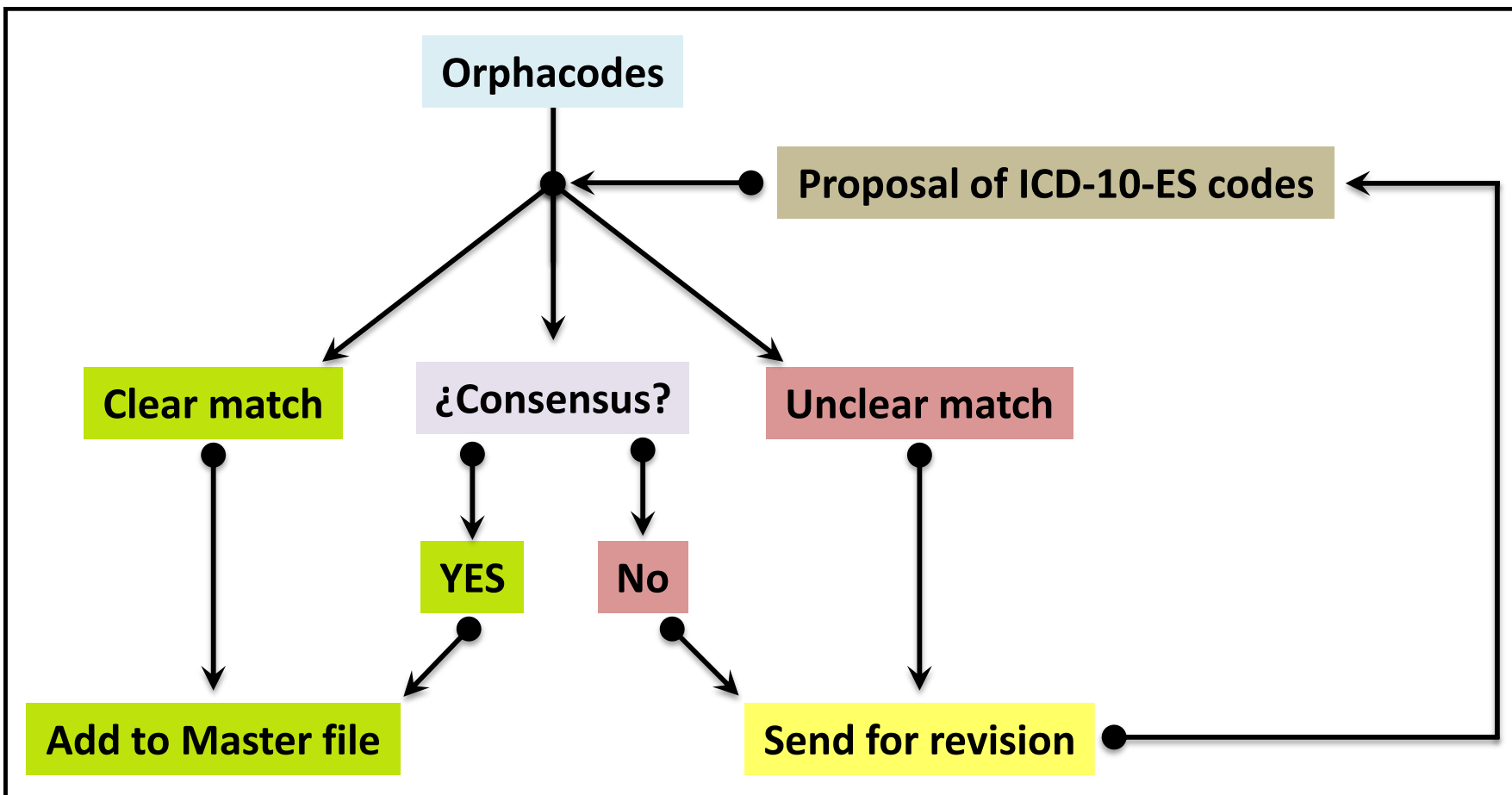
"Rare diseases are rare, but rare disease patients are numerous"

Access our Services

- Inventory, classification and nomenclature of rare diseases, with genes involved
- Inventory of orphan drugs
- Directory of patient organisations
- Directory of professional and institutions
- Directory of expert centres
- Directory of medical laboratories providing diagnostic tests
- Directory of ongoing research projects, clinical trials, registries and biobanks
- Collection of thematic reports Orphanet Research Series

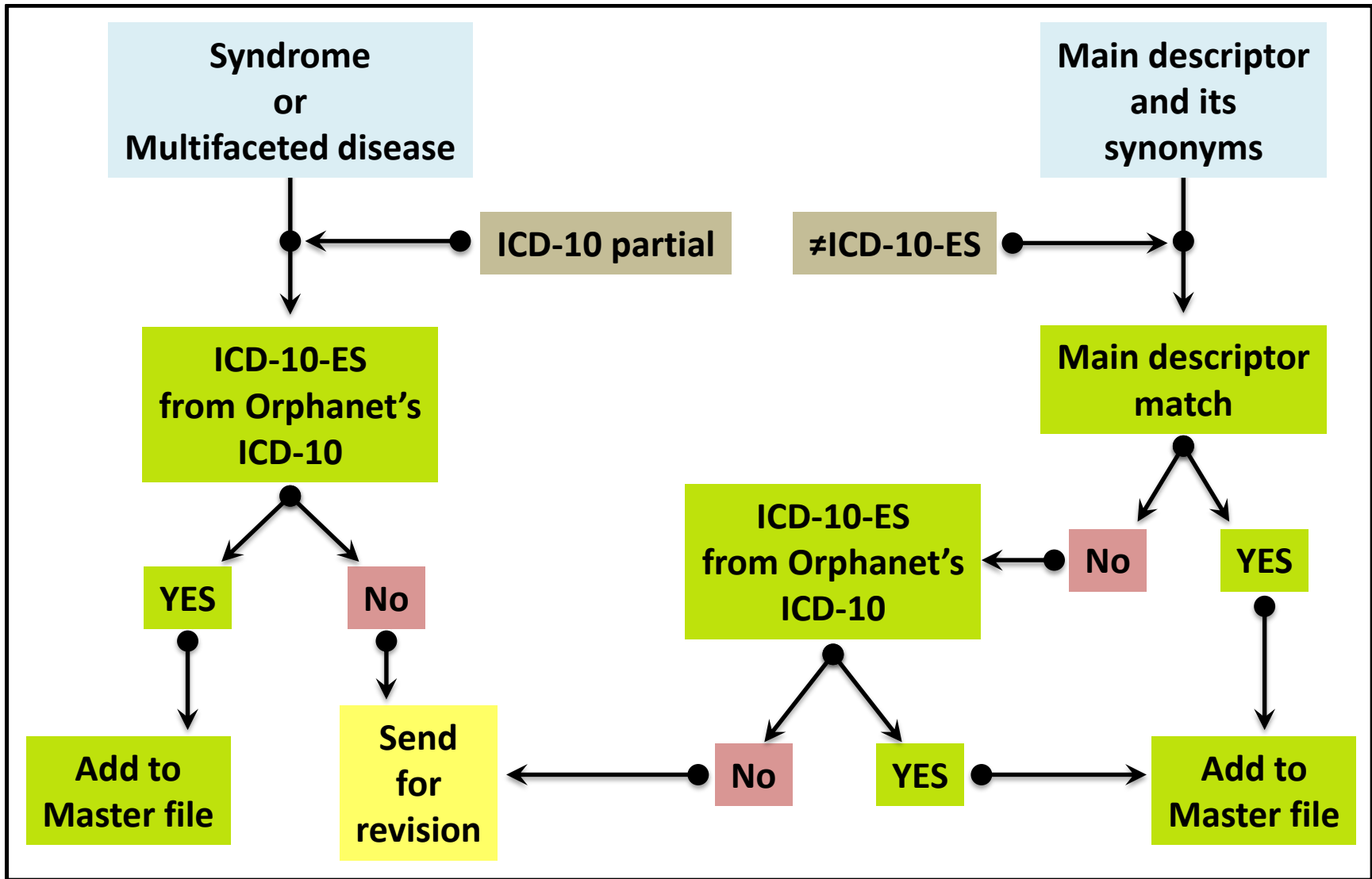


# Mapping ORPHAcodes to ICD-10-ES



1. Choose the ICD-10-ES code that has an exact match for the name of the disease.
2. Prioritize the ICD-10 (from Orphanet) derived ICD-10-ES code.
3. Send for revision the equivalences that remain unclear.

# Decision-making criteria



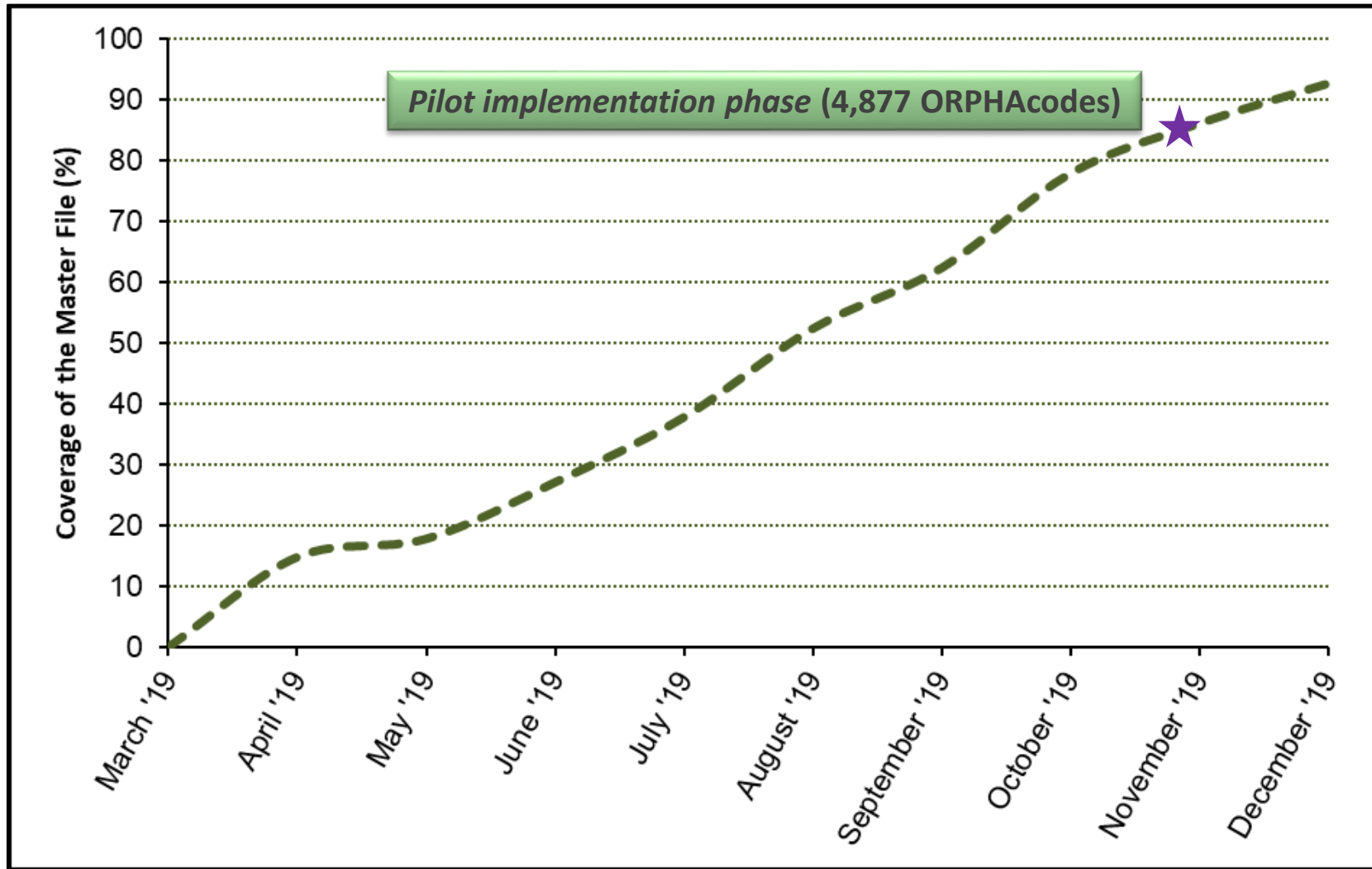
# Syndrome or Multifaceted disease

ORPHA	ORPHA Name	ICD-10 V2016	ICD-10-ES 2019	ICD-10-ES Name
163	Hereditary hyperferritinemia-cataract syndrome	H26.0	<b>E83.19</b> H26.0*	<b>Otros trastornos del metabolismo del hierro</b> Catarata infantil y juvenil
86815	Aplasia of lacrimal and salivary glands	Q10.4	Q10.4	Ausencia congénita de punto lagrimal
		Q38.4	Q38.4	Ausencia congénita de las glándulas y de los conductos salivales

# Main descriptor and synonyms

ORPHA	ORPHA Name	ICD-10 V2016	ICD-10-ES 2019	ICD-10-ES Name
1243	Best vitelliform macular dystrophy	H35.5	H35.54	Distrofia retiniana viteliforme
	Early-onset vitelliform macular dystrophy			
	Best disease		H35.50	Best, enfermedad de H35.50
86879	Extranodal nasal NK/T cell lymphoma	C86.0	C86.0	Linfoma extraganglionar de células NK/T, tipo nasal
	Lethal midline granuloma		M31.2	Granuloma letal de línea media

# Coverage of the Master file



- ❖ 2,102 different ICD-10-ES codes were proposed to 5,232 ORPHAcodes
- ❖ 543 ORPHAcodes remain under revision

Results gathered from the regional registries

# PILOT IMPLEMENTATION PHASE

# Castile and Leon

## > Information sources:

- ★ Minimum basic data set (CMBD)\*
- ★ Primary care\*
- ★ Mortality registry
- ★ Regional renal diseases registry
- ★ Early-detection congenital diseases registry
- ★ Rare diseases diagnosis centre (DierCyL)

\*First two provide 98% of the data

# Castile and Leon

## > Main goals 2019:

- ★ Change reference coding system to ICD-10
- ★ Test implementation options of ORPHAcodes

## > Outputs 2019:

- ★ Setup of new software version with ICD-10
- ★ Characterization of cases registered
- ★ Inclusion of the ORPHAcode variable
- ★ Translation of diagnoses to ORPHAcodes



# Catalonia

## > Information sources:

- ★ Clinicians from selected units of clinical experience networks in line with European Reference Networks
- ★ Minimum basic data set (CMBD)
- ★ Regional renal disease registry
- ★ Neonatal screening
- ★ Drugs registry

# Catalonia

## > Main goals 2019:

- ★ Create a working group of clinical health documentation professionals
- ★ Promote the use of correspondences in Catalonia to facilitate the retrieval of RD epidemiological data

## > Outputs 2019:

- ★ Inclusion of the Master file at the Catalan Health Department terminological server
- ★ Inclusion of the ORPHAcode database
- ❖ Out of >850 common ORPHAcodes between the MF and the registry:
  - ✓ 671 had the same ICD-10-ES equivalence
  - ✓ 64 had different equivalence
  - ✓ 138 had additional equivalences

# Murcia

## > Information sources:

- ★ Minimum basic data set (CMBD)
- ★ Medical records
- ★ Renal diseases registry
- ★ Dependency registry
- ★ Drugs registry
- ★ Patient referral registry
- ★ Disability registry
- Not limited to those mentioned above

# Murcia

## > Main goals 2019:

- ★ Filter the MF to exclude the non-considered
- ★ Assign ORPHAcodes to retrieved ICD-10-ES
- ★ Test the functionality of the system

## > Outputs 2019:

- ★ 30% of the cases validated as RDs
- ★ 70 ICD-10-ES → 158 cases
- ★ 90% of the cases linked to MF ORPHAcodes

# Valencian Region

## > Information sources:

- ★ Minimum basic data set (CMBD)
- ★ Congenital anomalies registry
- ★ Mortality registry
- ★ Neonatal screening
- ★ Renal diseases registry

# Valencian Region

## > Main goals 2019:

- ★ Integrate ICD-10-ES to ORPHAcodes matches
- ★ Automate ORPHAcodes assignment

## > Outputs 2019:

- ★ 1 ICD-10-ES → Many ORPHAcodes
  - Hierarchical ORPHAcodes selection
  - Creation of virtual ORPHAcodes
  - Manually revised and confirmed/changed
- ★ Many ICD-10-ES → 1 ORPHAcodes
  - The original ICD-10-ES is kept

# Navarre

## > Information sources:

- ★ Minimum basic data set (CMBD)
- ★ Primary care
- ★ Mortality registry
- ★ Temporal disability registry
- ★ Drugs registry
- ★ Congenital anomalies registry
- ★ Genetics service

# Navarre

## > Main goals 2019:

- ★ To create a working group of healthcare information systems professionals to promote the use of Orphacodes
- ★ To implement Orphacodes descriptors and synonyms

## > Outputs 2019:

- ★ Software preparation to allocate synonyms list
- ★ Initial contacts with Navarre Health Service to implement MF in medical records management



# Basque Country

## > Information sources:

- ★ Direct notification by Physicians

## > Main goal 2019:

- ★ Develop the Master file as a useful tool for the Basque Country Health service – Osakidetza

## > Outputs 2019:

- ★ Active collaboration to reach ICD-10-ES to ORPHA
- ★ Contacts with Osakidetza to include the MF within the medical records management tool

Strengths and weaknesses

# SUMMARY & CONCLUSIONS

# Implementation of ORPHAcodes

	Adaptations towards ORPHAcodes	ORPHAcodes implemented
Basque Country	x	x
Catalonia	x	x
Castile and Leon	x	x
Murcia	x	x
Navarre	x	
Valencian Region	x	x

# Regional registries working features

## > ICD based information sources:

- ★ Direct impact over ORPHAcodes assignment
- ★ MF sets the criteria to unify ORPHAcodes

## > Direct notification by Physicians

- ★ Partial impact over ORPHAcode assignment
- ★ MF facilitates communication among services

# Mapping of ICD to ORPHAcodes

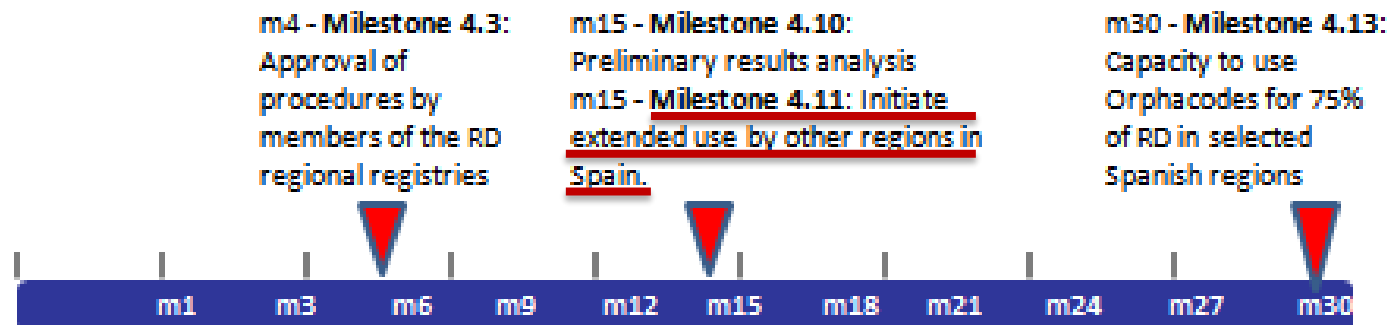
## > Direct Translation:

- ★ There is about 1 ICD-10-ES for 3 ORPHAcodes
- ★ Almost 1 in 10 ORPHAcodes have no ICD-10 proposed from Orphanet

## > ICD-10-ES redundancy:

- ★ The lack of specific ICD-10-ES codes leads to the clustering of ORPHAcodes
- ★ These redundancy makes unlikely to automate the process and tedious to validate the equivalences

# Timeline



## Phase 1

**Pilot phase:** Use of the “Standard procedure and guide for the coding with Orphacodes”  
And the “Specification and implementation manual of the Master file” by a selected group of regions



## Phase 2

► m15 – Deliverable 4.4: Preliminary results analysis

## Initiated

## Phase 3

**Extended** use of the “Standard procedure and guide” and  
“Specification and implementation manual of the Master file”  
by other regions / national level based  
on the results obtained in Phase 1

## Phase 4



m30 – Deliverable 4.11: Final Report  
including **Recommendations** for implementation  
at a national level

More codes, more registries

# THE FOLLOW-ON PROCESS

# Completing and updating the MF

## > Consensus based equivalences:

### ★ 118 unclear matches

- 81 solved by agreement among registries
- 37 remain under revision

## > ORPHAcodes without ICD-10-ES proposed:

### ★ 404 out of 425 didn't have ICD-10 from Orphanet

- 18 were found to have updated ICD-10 information
- 205 were assigned an ICD-10-ES equivalence
- 220 remain under revision

## ❖ **5518 total ORPHAcodes mapped to ICD-10-ES**

- ✓ Updated version distributed in May 2020



# Expanding the regional network

## > Galicia:

- ★ Population → 2.7 million inhabitants
- ★ Coding systems in use → ICD-10, Snomed-CT
- ★ Information sources → Minimum Basic Data Set, Drugs registry, Neonatal screening-Metabolopathies, Mortality, Renal diseases, active search, and others
- ★ Number of cases → 1,900 cases confirmed for the period January 2018 – June 2020.
- ★ Validation protocol → reviewed using Medical Records

# Expanding the regional network

## > La Rioja:

- ★ Population → 317,000 inhabitants
- ★ Coding systems in use → ICD-10, ICD-10-ES
- ★ Information sources → Minimum Basic Data Set, Mortality registry, Genetics laboratory
- ★ Number of cases → 11,763 patients since 2010.
- ★ Validation protocol → Manually curated

# Expanding the regional network

## > Madrid:

- ★ Population → 6.6 million inhabitants
- ★ Coding systems in use → ICD-10, ICD-10-ES, ERA-EDTA
- ★ Information sources → Minimum Basic Data Set, Primary Care, Renal diseases, Neonatal screening, Orphan drugs, mortality, and direct notification
- ★ Number of cases → 350,000 patients (17,000 confirmed cases) for 2010-2017 period
- ★ Validation protocol → Automated and manual

# Expanding the regional network

- > Catalonia (7.5 M)
- > Basque country (2.2 M)
- > Navarre (640 K)
- > Castile and Leon (2.5 M)
- > Valencian Region (5 M)
- > Murcia (1.5 M)
- > Galicia (2.7 M)
- > La Rioja (320 K)
- > Madrid (6.6 M)

*Almost*

**29 million**

*People*

*Around*

**60%**

*of the Spanish population*

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Time to discuss

# THANK YOU!

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*ciberer*



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