Use-Cases ORPHAcodes Implementation in Germany, France, the Netherlands & Norway
June 2024

Use cases of ORPHAcodes Implementation (table 1 RD diagnosis coding, table 2 Additional info/specific Use cases and table 3 Data Exploitation)

Table 1. RD diagnosis coding

Country	Where	Inpatient/Outpatient	Registration of diagnosis	Who codes	Coding diagnosis: file used	Number of OC available	Visibility of the OC or RD diagnosis in the EHR (further encounter)
🇫🇷	Mandatory for ALL RD centres in Hospitals	Both	Tab RD available in the EHR and daily transmission to intermediate system (BAMARA) (after anonymization) which feeds specific registries and the National RD registry (BNDMR) and other data repository and eprescription.	Clinicians/ARC from expert centres (with specific funding to increase coding capacity) type a diagnosis and the OC is automatically retrieved.	List of diagnosis names linked to OC/ Nom Pack/ FLAT no classification).	As per Nom Pack	Yes the code follows the patient
🇩🇪	Mandatory for ALL Hospitals	Inpatient	Clinicians provide a diagnosis	Clinicians register the diagnosis and then Coders or Clinicians assign a code	Alpha-ID-SE file as basis for coding tools of the hospital software manufacturers. The ORPHAcodes are aligned to ICD10-GM by the Orphanet DE/BfArM experts	7,022 If no ICD10 term => no OC	Yes (if EHR exists, not mandatory yet)
🇳🇱	ALL Hospitals using the Diagnosis Thesaurus (DT)	Both	Diagnosis Thesaurus (DT) is used to register diagnosis in EHR. The EHR feeds the DHD ORPHA-viewer.	Clinicians register the diagnosis and the linked ORPHAcode is automatically retrieved.	Mapping file aligns the Diagnosis Thesaurus (DT - originally based on a subset of SNOMED-CT) with SNOMED-CT and ORPHAcodes.	Diagnosis Thesaurus (DT) currently contains 5,500 diagnoses with a linked ORPHAcode	Specific field for the RD thesaurus name (field for OC in development).
🇳🇴	All hospitals in one of 4 health regions (covering approx. Half of NO inhabitants)	Both	EHR and feeds the National registry (manually)	Clinicians in expert centres (specialized health care service) and then the code follows the patient	Clinicians enter the OC in dedicated RD tab of the EHR (from ORPHAcodes API and Orphadata API (classifications and associated genes)	As per Nom Pack	An icon appears next to the patient name whenever an OC has been attributed

Table 2 Additional Info/ use cases

Country	Diagnosis status options available	Additional descriptors	Inactivated codes	Undiagnosed code	Group codes allowed	Non Chronic RD history shown?
France	Ongoing, Probable, confirmed, undetermined	ICD10, HPO, Genes HGNC, Atypical signs, OC (groups allowed)	Removed or referred to with each new issue. Not retroactively on already registered diagnosis, a QC is carried out whenever data is exploited at the registry level to remove all the inactivated codes.	Available	Yes for "ongoing" diagnosis AND in the Additional information box	No
Germany	Not available	Not available	Removed from the file upon Annual Release of Nom Pack (Differential File)	Available	Yes some are present in the file	No
Netherlands	Not available: Doctors register (working) diagnosis and should adjust when more information is collected during the process.	Thesaurus-ID, SNOMED and ICD-10 codes	Because the clinicians register with diagnosis thesaurus, this ID stays the same and the code linked to it is updated automatically with each new pack	Not available	Yes	Yes, in medical history
Norway	Not available. OC inserted ONLY when all investigations have been carried out = definitive diagnosis.	ICD-10	When accessing the patient file, the clinician is notified and requested to update the OC	Available	Yes. However, when a group code is chosen, a warning notification pops up, recommending using a code at disorder or subtype level for confirmed diagnosis	Yes: dedicated box indicates Diseases & RD that are cured

Table 3. Data Exploitation

Country	Exploitation tool	OC File used for exploitation	Quality Control of registered data	Numbers, reports, analysis & links reference documents available
(France flag)	Anonymized data in the National RD registry system n (BNDMR/pyramig). Pyramig allows to provide statistics by NRN centre. They exploit the xml files including the classifications RD Can be linked to other repository i.e système national de données de santé (SNDS), for socio-economical studies	Nom Pack including classifications	Yes by the National RD Registry_BNDMR staff. Liaise with clinicians if needed (coherent code, inactivations….)	1,372 M patient registered in the National RD Registry BNDMR & several publications and analysis https://www.bndmr.fr/publications/nombre-de-cas-par-mr/
(Germany flag)	Alpha-ID-SE file, ORPHAcodes based on Orphanet nomenclature pack	Alpha-ID-SE file, ORPHAcodes based on partial Orphanet nomenclature pack Flat file	No	Not available
(Netherlands flag)	ORPHA-viewer: it exploits data registered in the EHR. Future developments: the ability to use the tool for research purposes, policy making, data exchange with ERNs and the national designation of expert centres.	The tool Exploits the ORPHAcodes API and can aggregate data and exploit the classification.	No	Not yet available
(Norway flag)	RD REGISTRY	Nom Pack (Flat file without classifications. Aggregation Level is included)	No	3,900 patients registered in the RD Registry (as of June 2024). Annual report for 2022 available in Norwegian arsrapport-sjeldenregisteret-2022-endelig.pdf (oslo-universitetssykehus.no)