

ORPHAcoding RD Patients in a Rare Disease Centre - The Tübingen Solution

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The Centre for Rare Disease at the University Hospital Tuebingen (CRDT) has implemented ORPHAcoding into the local hospital information system (SAP), thus enabling identification and counts of rare disease (RD) patients. Since 2019 all inpatient and outpatient rare disease patients seen at CRDT are being ORPHAcoded. The distribution of the healthcare funding within the CRDT subunits is based upon numbers of ORPHAcoded RD patients. These RD patient counts are also reported to the healthcare authorities and health insurances for recognition and funding of the CRDT, and furthermore, will be utilized for the RD use case of the German Medical Informatics Initiative.

Composition of CRDT

The CRDT was set up in 2010 and is composed of two main building blocks. Firstly, the umbrella structure of the CRDT contains the coordination office, the academy for further medical training, the educational program, the treatment research centre and the care pathway for undiagnosed patients. Secondly, 14 CRDT subunits, which are care and research centres themselves, focus on specific RD diseases and/or disease groups. Cases in point are the Centre for Rare Neurological Diseases (ZSNE), the Autoinflammation Reference Centre (ArcT), the Centre for Rare Liver Diseases (ZSL), etc. (www.zse-tuebingen.de for complete overview)

Implementing ORPHAcoding at CRDT

The CRDT has access to two options to ORPHAcode: (i) The CRDT subunits use custom-made ORPHAcode lists that reflect their respective expertise. (ii) The entire hospital can use the diagnostic coding software ID DIACOS to ORPHAcode.

Furthermore, to ensure consistency and to avoid extra work, for patients that have already been ORPHAcoded in the past, the respective ORPHAcode(s) can easily be re-entered using the patient monitoring tool within the hospital information system SAP is-h.

CRDT medical staff and coding specialists are offered training courses in ORPHAcoding, and regular information about ORPHAcoding is now part of the periodical meetings for hospital coding specialists.



Fig. 1: Accessing ORPHAcodes

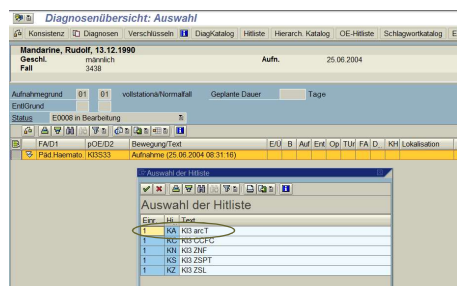


Fig. 2: Choice of ORPHAcode lists for CRDT pediatricians in is-h, arcT at top

ID DIACOS for the entire hospital

It is desirable that all hospital RD patients are registered using ORPHAcodes, not only CRDT patients. Apart from the safety benefit that a RD patient can be easily identified if hospitalised, the medical controlling unit may use ORPHAcodes for ambulatory care-sensitive hospitalisation purposes. In addition for CRDT recognition the entire hospitals needs to show a given number of RD inpatients, and it is difficult to impossible obtaining accurate RD patient counts using icd-10-codes.

ID DIACOS (diagnostic coding software) contains the DIMDI (German Institute of Medical Documentation and Information) alpha-id-se-test file that links ORPHAcode and ICD-10-GM-code. However, the alpha-id-se-file does not yet contain all rare diseases, and predominantly on level disorder, making custom-made ORPHAcode lists essential for the CRDT.

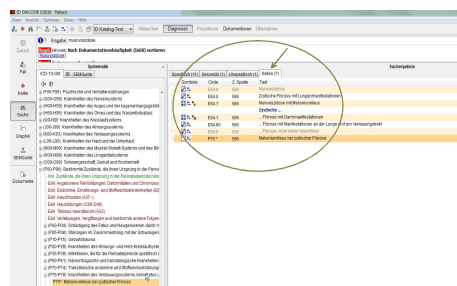


Figure 5: ID Diacos, enabling ORPHAcoding RD patients using the position „Selten“

Results

- Two ways of ORPHAcoding implemented at CRDT and University Hospital Tübingen: (i) custom-made ORPHAcode lists for CRDT subunits and (ii) all ORPHAcodes via the diagnostic coding software ID DIACOS
- ORPHAcode lists were implemented for 14 CRDT subunits
- Total numbers of ORPHAcodes contained at CRDT subunit lists: 931 (by 31.12.2019)
- ORPHAcoded patients and cases at CRDT in 2019 (cut-off date 04.02.20)
 - outpatient patients: 4.225, outpatient cases: 6.015
 - inpatient patients: 1.211, inpatient cases: 2.043

Discussion

To our knowledge, the CRDT is the first Rare Disease Centre (RDC) that has implemented an *expertise* reflecting ORPHAcoding. In particular, for rare disease care it is important that an expertise claim is confirmed by a number of patients seen of a certain disease or disease group. Expertise of a RDC is being determined in Germany according to a catalogue of conditions and criteria that a RDC has to fulfil which has been defined by the German National Action Plan for Rare Diseases.

By using ORPHAcoding in the CRDT, we now know how many patients with a certain RD have been seen. Introducing ORPHAcoding does come with extra work, however, it is the only reliable source for identification of RD patients and for counting patient numbers and cases. A method of permanently marking RD patients with a confirmed diagnosis within the local hospital information system is under way.

Implementation of ORPHAcoding at CRDT was initially driven by the intention to have a reliable source for assessing performance and development of the centre, however, it has become a formal requirement in Germany as laid down in the centre regulation by the Federal Joint Committee decision ('GBA-Beschluss').

Introducing ORPHAcoding at the CRDT will, furthermore, provide the needed interoperable interface to the registries all European Reference Networks (ERN) are currently establishing or have established yet.

In addition, since 2020 there is a RD use case (CORD) of the German Medical Informatics Initiative underway which will be collecting aggregate numbers of rare disease patients seen at all German Rare Disease centres. The very basis for being able to collect these numbers is the availability of ORPHAcoded patients.

Acknowledgements

Tobias König (former UKT employee, for customising Qlik View to CRDT requirements), Karl-Eugen Schairer (former IT UKT, for implementation of ORPHAcode lists into SAP), Kurt Kirch (DIMDI, for adding CRDT required ORPHAcodes to the DIMDI alpha-id-se-list whenever possible and answering many questions; <https://www.dimdi.de/dynamic/de/klassifikationen/icd/alpha-id/nationales-projekt-kodierung-se/>), Kathrin Rommel (Orphanet Germany, for helping obtaining ORPHAcodes for disorders that have yet not been given an ORPHAcode; <http://www.orpha.net/national/DE-DE/index/team-kontakt/>).

Custom-made, expertise reflecting ORPHAcode lists for CRDT subunits

All CRDT subunits put together ORPHAcode lists that reflect their expertise and the patients they see. Selected ORPHAcodes are whenever possible on the specificity level *disorder* and thus in accordance with the alpha-id-se-list provided by DIMDI (German Institute of Medical Documentation and Information). The alpha-id-se-list links ORPHAcode and ICD-10-GM-code via so called disorder synonyme specific alpha-id's.

Medical controlling and IT staff uploaded these ORPHAcode lists into the local hospital information system.

The CRDT subunits have exclusive access to their individual ORPHAcode lists, hence ensuring that all needed ORPHAcodes are handily and completely available. ORPHAcode lists are only available for coding within the respective coding environment of CRDT subunits, not hospital-wide.

The roll-out of these lists was and is supported by customised training courses. CRDT staff is advised to only use ORPHAcodes from their own ORPHAcode lists as these are the basis for performance evaluation. This prevents using alternate ORPHAcodes (e.g. by using DIACOS) that would not be considered when analysing the CRDT data.

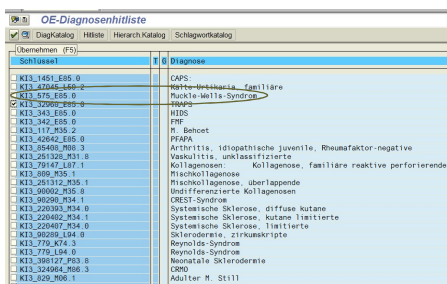


Figure 3: ORPHAcode list for CRDT subunit arcT in is-h, showing coding environment, ORPHAcode, icd-10-code, name of disorder Muckle-Wells-Syndrom encircled

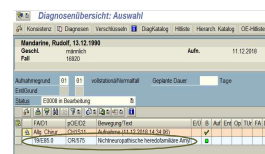


Figure 4: Patient with RD coded with ORPHAcode and icd-10-code, name of disorder as given by icd-10-code.

Counting RD patients using ORPHAcodes and QlikView

QlikView (data analysis software) allows to count patient and case numbers within CRDT subunits and hospital-wide.

Moreover, QlikView can be customised to the individual CRDT subunits and thus provides a quick and efficient way to analyse CRDT numbers. The analysis is based on two parameters (i) the CRDT subunit ORPHAcode list and (ii) the respective organisational units at which the seen patients are documented in the hospital management system. Using these two parameters for CRDT counts makes sure that RD patients are only counted when seen within the CRDT, and not for (potential) other medical treatment at the hospital.

Diagnosedatum:		Anzahl Patienten nach Zentrum mit PDE	
Jahr	Quartal	ORPHAcode	ORPHAcode_Bezeichnung
2019	1	92	Arthritis, idiopathische juvenile
2019	2	117	Behçet-Syndrom
2019	3	342	Mittelmerefebr, familiäres
2019	4	243	Hypertonie/globuläre D mit Rückfallrisiko
2019	1	575	Muckle-Wells-Syndrom
2019	2	703	Periostitis, bulloides
2019	3	515	CREST-Syndrom
2019	4	2331	Kawasaki-Krankheit
2019	1	3287	Takayasu-Arteritis
2019	2	32960	Tumormarkierungsfaktor-Rezeptor 1-assoziiertes p.
2019	3	42643	Behçet-Syndrom
2019	4	47045	Kälte-urikaria, familiäre
2019	1	85418	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85419	Arthritis, idiopathische juvenile, oligoartikuläre
2019	3	85420	Arthritis, idiopathische juvenile, Entzündung
2019	4	85421	Arthritis, idiopathische juvenile, systemische
2019	1	85422	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85423	Arthritis, idiopathische juvenile, Rheumafaktor
2019	3	85424	Arthritis, idiopathische juvenile, Rheumafaktor
2019	4	85425	Arthritis, idiopathische juvenile, Rheumafaktor
2019	1	85426	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85427	Arthritis, idiopathische juvenile, Rheumafaktor
2019	3	85428	Arthritis, idiopathische juvenile, Rheumafaktor
2019	4	85429	Arthritis, idiopathische juvenile, Rheumafaktor
2019	1	85430	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85431	Arthritis, idiopathische juvenile, Rheumafaktor
2019	3	85432	Arthritis, idiopathische juvenile, Rheumafaktor
2019	4	85433	Arthritis, idiopathische juvenile, Rheumafaktor
2019	1	85434	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85435	Arthritis, idiopathische juvenile, Rheumafaktor
2019	3	85436	Arthritis, idiopathische juvenile, Rheumafaktor
2019	4	85437	Arthritis, idiopathische juvenile, Rheumafaktor
2019	1	85438	Arthritis, idiopathische juvenile, Rheumafaktor
2019	2	85439	Arthritis, idiopathische juvenile, Rheumafaktor
2019	3	85440	Arthritis, idiopathische juvenile, Rheumafaktor
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2019	3	85444	Arthritis, idiopathische juvenile, Rheumafaktor
2019	4	85445	Arthritis, idiopathische juvenile, Rheumafaktor
2019	1	85446	Arthritis, idiopathische juvenile, Rheumafaktor
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