D 4.10 Romania Outcome Report

REPORT

December 2021

This report is part of the project 826607 RD-CODE which has received funding from the European Union's Health Programme (2014-2020).

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Background

In Romania (population of 20 million inhabitants) there is no ORPHAcode implementation in routine coding systems at national level. The health system uses DRG coding and ICD10 for reporting rare disorders. Most of the cases are reported by the 6 Regional Medical Genetics Centres (Bihor, Bucharest, Cluj, Dolj, Iasi, Timis) nominated in 2014. Iasi Regional Medical Genetics Centre covers 9 districts (Moldova province) that accounts approximately for 20% of the Romanian population. Congenital anomalies and genetic disorders were systematically reported until 1989. After 1989, there is no national inventory of rare disorders, only Bihor Regional Medical Genetics Centre contributed to EUROCAT and thus has a registry in the field.

Objectives

To introduce ORPHAcodes, according to the « Standard procedure and guide for the coding with Orphacodes » and the « Specification and implementation manual of the Master file » in daily practice of the Romanian Health System and to facilitate allocation of proper resources for patients with rare diseases. The work has started in Iasi center and later will be extended to other centres involved in ERNs.

Team description

Dr. Liviu Stafie - the manager of the Department of Public Health Iasi (Directia de Sanatate Publica Iasi) has discussed with the Health Minister representatives and has presented the benefits of Orphacodes for Health Information Systems and the possibilities to implement Orphacodes. The Health Ministry recently asked for specific numbers concerning rare diseases, but as the services do not have a specific information system working, this was a very difficult task. Thus, the introduction of Orphacodes in daily practice of the Health Information System will allow accurate reporting of RD cases.

Prof. Cristina Rusu - coordinator the Iasi Regional Medical Genetics Centre belonging to Iasi Children's Hospital and member of the National Council of Rare Diseases disseminated information about Orphacodes and Standard operating procedures at different conferences (Conference of the Romanian Society of Medical Genetics 2019; Iasi Pediatric Days 2019; Moinesti Hospital Days 2019; First National Congress of the Romanian Society of Audiology and Pathology of Communication 2019). She discussed with representatives from 6 Regional Medical Genetics Centres (Bihor, Bucharest, Cluj, Dolj, Iasi, Timis) and coordinator of National Alliance of Rare Diseases - Dorica Dan, in order to elaborate a work plan to introduce Orphacodes for each centre. Most of the rare diseases cases in the country are reported by the 6 Regional Medical Genetics Centres (Bihor, Bucharest, Cluj, Dolj, Iasi, Timis) nominated in 2014.

Senior lecturer Monica Cristina Panzaru MD PhD – a medical geneticist involved in the diagnosis of rare diseases patients – disseminated information about Orphacodes and rare diseases to colleagues from other medical specialities.

Activities carried out /achievements

Communication activities

1) Senior lecturer Monica Cristina Panzaru had a presentation “Coding of rare diseases with Orphacodes” at International Symposium “Teaching and learning innovations in medical education”.

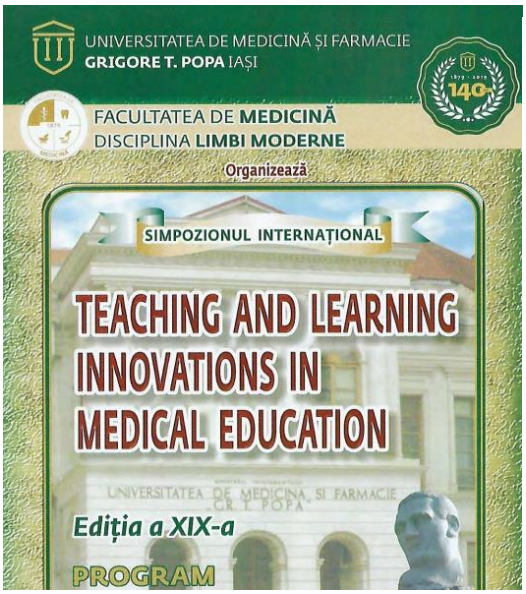
2) *Conference of the Romanian Society of Medical Genetics 2019*

3) *First National Congress of the Romanian Society of Audiology and Pathology of Communication*

Moinesti Hospital Days 2019

Conference of the Romanian Society of Medical Genetics 2019

First National Congress of the Romanian Society of Audiology and Pathology of Communication



10.00-15.00 SECȚIUNEA II –MEDICINE (Centrul de limbi moderne și integrare culturală – Amfiteatrul Mic)

Moderatori:
Cristian Stătescu (Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)
Marius Valeriu Hînganu (Universitatea de Medicină și Farmacie "Gr. T. Popa" Iași)
Radu Andy Sascău (Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)

Secretar:
Asist. Dr. Setalia Popa (Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)

1. Monica Pânzaru, Lavinia Caba, Lăcrămioara Ionela Butnariu, Elena Braha (*Institutul Național de Endocrinologie "C.I. Parhon", București*), Roxana Popescu, Setalia Popa, Irina Resmeriță, Eusebiu Vlad Gorduza, Cristina Rusu *(Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)* – **Coding of rare diseases with Orphacodes**
2. Monica Pânzaru, Cristina Rusu, Lavinia Caba, Lăcrămioara Ionela Butnariu, Mihaela Grămescu, Setalia Popa, Irina Resmeriță, Eusebiu Vlad Gorduza *(Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)* – **Genetic aspects of autism spectrum disorders**
3. Lăcrămioara Ionela Butnariu, Cristina Rusu, Monica Pânzaru, Lavinia Caba, Roxana Popescu, Setalia Popa, Irina Resmeriță, Cristina Rusu *(Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)* – **Probleme etice și genetice în tulburările de reproducere umană**
4. Lăcrămioara Ionela Butnariu, Cristina Rusu, Monica Pânzaru, Lavinia Caba, Roxana Popescu, Setalia Popa, Irina Resmeriță, Cristina Rusu *(Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)* – **Eterogenitatea genetică în sindroame cu acrocefalo-polisindactilie**
5. Irina Resmeriță, Sebastian Cozma, Oana Bitere, Roxana Popescu, Lavinia Caba, Monica Pânzaru, Lăcrămioara Butnariu, Setalia Popa, Luminița Rădulescu, Eusebiu Vlad Gorduza, Cristina Rusu *(Universitatea de Medicina si Farmacie "Gr. T. Popa" Iasi)* – **Hipoacuzia sindromică – dificultăți de diagnostic, management și tratament**

4) Prof. Cristina Rusu had a presentation « RD CODE : the importance of uniform codification of rare diseases » at Europlan online Conference, 26.11.2021.

AGENDĂ
CONFERINȚA EUROPLAN ONLINE
25 – 26 noiembrie 2021

Ziua 1	*25.11.2021*
14.00 -14.30	*Deschidere oficială EUROPLAN*
14.30-14.45	Progrese & provocări în domeniul bolilor rare – Advocacy la nivel național și internațional. Reprezentanți ai autorităților, Dorica Dan -președinte ANBRaRo;
14.45 -15.45	**Acces la diagnostic si tratament, moderator Prof. Dr. Maria Puiu, UMFT;**
14.45-15.00	1. Prezentarea Documentului: **ROMÂNIA – CĂLĂTORIA CĂTRE "RARE 2030"**, Obiectiv: Managementul integrat al pacientului cu boli rare, document realizat în parteneriat de către: ANBRaRo, CNBR, SRGM; Orphanet și Takeda – Prof. Dr. Maria Puiu, UMFT
15.00-15.15	2. **"Dreptul la diagnostic - Utilitatea screeningului metabolic neonatal extins in identificarea maladiilor metabolice ereditare din perspectiva geneticii medicale"** , Dr. Plăiașu Vasilica, CRGM București, INSMC Alessandrescu-Rusescu;
15.15-15.30	3. **"Boli rare ereditare renale: provocări și noi perspective"**, Dr. Elena Rusu, Institutul Clinic Fundeni
15.30-15.45	4. **"Testarea genomică în bolile rare"**, Prof. dr. Jurca Claudia, Prof. dr. Bembea Marius, Universitatea din Oradea, Facultatea de Medicină și Farmacie Spitalul Clinic Municipal, dr. Gavril Curteanu" Oradea, Centrul Regional de Genetică Medicală Bihor
15.45 – 17.00	**Raport „Monitorare" – Monitorizarea implementării PNBR în România, Prof. Dr. Emilia Severin, UMFB. Dezbatere.**
Ziua 2	*26.11.2021*
14.00 – 15.30	**Boli rare: Îngrijirea pacienților cu boli rare, moderator dr. Violeta Stan**
14.00-14.15	1. **Provocări in îngrijirea pacienților cu boli reumatologice rare in 2021**, Dr. Laura Damian, Centru de Expertiză pentru boli musculo scheletale rare.
14.15-14.30	2. **Centrul de Boli Rare Oculare Timișoara – noi începuturi**, Florina Stoica, Alexandra Ionescue
14.30-14.45	3. **Angioedemul Ereditar in Romania: situația actuală privind accesul la tratament**, Dr. Bara Noemi, Centrul de Expertiză pentru Angioedem Ereditar Tg. Mureș
14.45-15.00	4. **Modificarea paradigmei legate de educația si cercetarea vizând bolile rare**, Conf. Dr. Nicoleta Andreescu, UMFT
15.00-15.15	5. **Rețeaua Ro-NMCA ID - participarea la ERN ITHACA- avantaje, limite, probleme, S.L. Dr. Adela Chiriță-Emandi, UMFT, Centrul Regional de Genetica Timiș, Centrul de Expertiză Ro-NMCA ID – ERN ITHACA**
15.15 – 15.30	6. **RD CODE – importanța codificării unitare a bolilor rare, Prof Dr. Cristina Rusu, UMFI, Orphanet România**
15.15-15.30	**Întrebări, comentarii**
15.30 -16.30	**Raport „Monitorare" – Monitorizarea implementării PNBR în România, Prof. Dr. Emilia Severin, UMFB. Dezbatere.**
16.30 – 17.00	**Concluzii & Rezoluția Europlan, Dorica Dan – președinte ANBRaRo**

5) Prof. Cristina Rusu had a presentation « CORE-RD : ORPHA codes for rare diseases in Romania » at Rare diseaseas School for journalists, 15.12.2021.

Alianța Națională pentru Boli Rare România

Agendă
Școala de Boli Rare pentru Jurnaliști
15.12.2021 – 16.12.2021

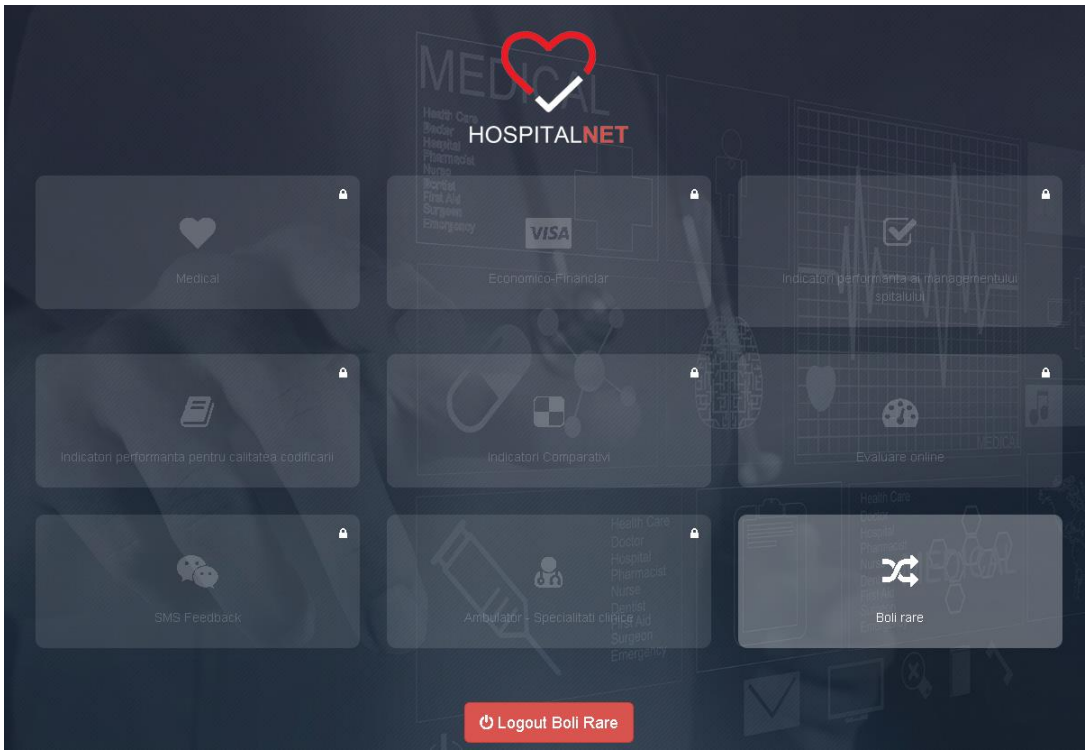
Miercuri	Sesiunea 2 - 15.12.2021, ora 14.00
14.00 - 14.10	Welcome **– Alexandra Mănăilă, jurnalist**
14.10 - 14.20	Implicare în sănătatea digitală – ANBRaRo 2021 **Dorica Dan, președinte ANBRaRo**
14.20 – 17.00	**Sesiune plenară**
14.20 – 14.50	**Lupta pentru cucerirea universului bolilor rare - genomica, terapii genice, terapii celulare,** Dr. Marius Geantă, Centrul de Inovare în medicină
14.50 – 15.00	**Q&A**
15.00 – 15.30	**Digital and Data Advisory Group in context European,** Veronica Popa – EURORDIS,
15.30– 16.00	**CORE – RD, codificarea ORPHA pentru bolile rare în România,** Prof. Dr. Cristina Rusu, UMF Iasi
16.00– 16.30	**Formare profesională in boli genetice și boli rare - ProGeneRare -** raport final implementare proiect, Ioana Streata, UMF Craiova
16.30 – 17.00	**Concurs Podcast Boli Rare – cine?**
	Concluzii

IT developments

According to the Standard operating procedures and specification and implementation manual an elaborate support was created for software developers to incorporate Orphacodes into Health Information Systems.

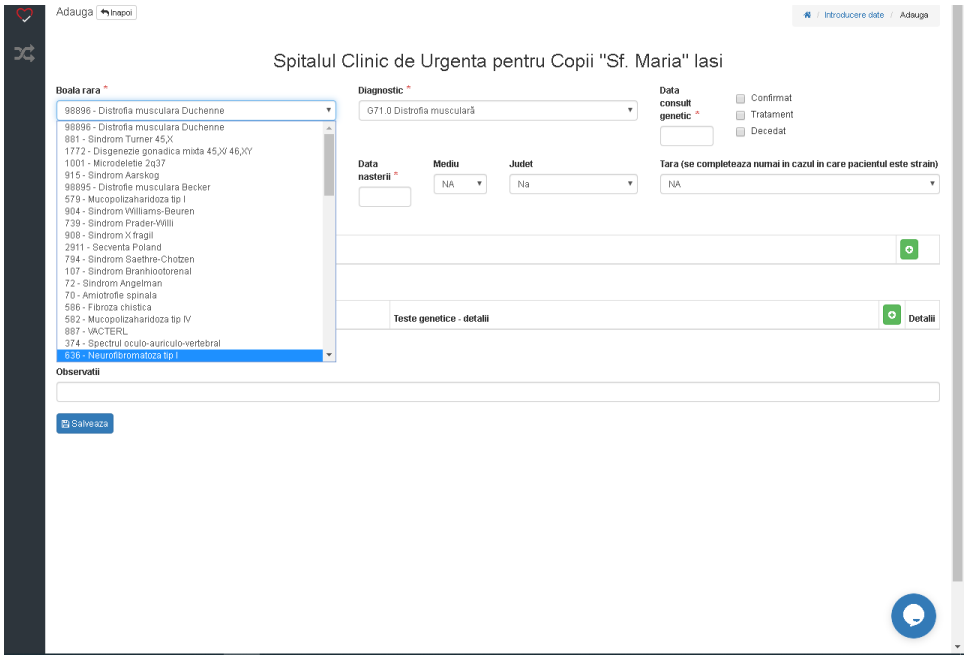
An IT tool was selected for ORPHAcode registration (fig. below).

A training on the IT tool was organized in order for the users to get acquainted with the tool.

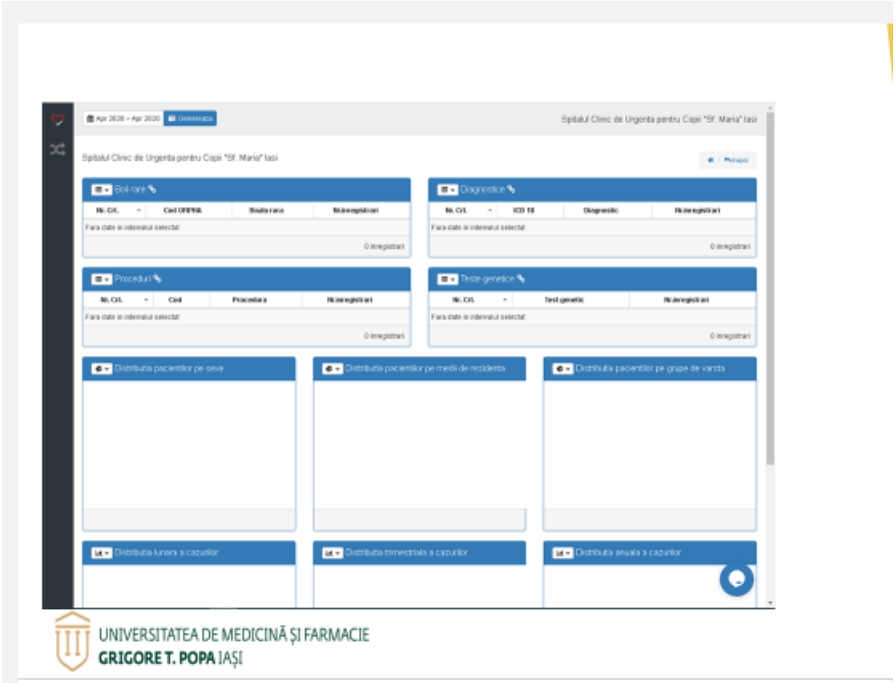


IT tool

It allows to choose between 193 different ORPHAcodes.



Clinicians can complete with additional information (fig below).



And the tool was designed as to allow to generate statistics (fig below).

Training activities

1) A training on ORPHAcodes for clinicians (Medical Genetics residents, Pediatricians) involved in Iasi Regional Medical Genetics was organized.

2) 3 Trainings for Timis, Nephrology Parhon Iasi and Zalau Centres were organized.

Ongoing work : RD registration

We started case registration in Iasi Regional Medical Genetics Centre). Iasi Regional Medical Genetics Centre covers 9 districts (Moldova province) that accounts approximately for 20% of the Romanian population.

A list of 193 rare diseases with associated ORPHAcodes was created in IT tool as described above.

A periodic evaluation of work is carried out by the implementing center. Difficulties were identified and discussed with IT team, National Coordinator and Project Manager.

3 additional centres in Zalau county started case registration in December 2021: C. I. Parhon Hospital Iasi – Nephrology Department, Timis Regional Medical Genetics Centre and Prader Willi Association - NoRo Expert Centre for Rare Diseases.


Introducere date

Introducere date / Boli rare

Spitalul Clinic de Urgenta pentru Copii "Sf. Maria" Iasi

Pe pagina: 10 Cauta:

ORPHA	Boala rara	Diagnostic ICD10	Proceduri	Data nasterii	CID	Data consult genetic
636	Neurofibromatoza tip I	Q85.0 Neurofibromatoza (nemaligna)	G01 MORFOMETRIE,G12 FOTOGRAFIERE,G04 EXTRACTIE ADN	2006-01-05	40187580510831928474	2019-12-03
636	Neurofibromatoza tip I	Q85.0 Neurofibromatoza (nemaligna)	G01 MORFOMETRIE,G12 FOTOGRAFIERE,G04 EXTRACTIE ADN	2005-03-12	40579842157046940602	2019-10-03
739	Sindrom Prader-Willi	Q87.14 Sindromul Prader-Willi	G01 MORFOMETRIE,G04 EXTRACTIE ADN,G15 MLPA P036/070 PENTRU SCREENING SUBTELOMERE,G12 FOTOGRAFIERE	2018-10-10	40656167906836534836	2019-04-04
739	Sindrom Prader-Willi	Q87.14 Sindromul Prader-Willi	G01 MORFOMETRIE,G12 FOTOGRAFIERE	2015-08-07	40148741222726131878	2019-04-23
739	Sindrom Prader-Willi	Q87.14 Sindromul Prader-Willi	G01 MORFOMETRIE,G12 FOTOGRAFIERE,G04 EXTRACTIE ADN,G15 MLPA P036/070 PENTRU SCREENING SUBTELOMERE	2015-06-08	40163163723064381686	2019-01-18
739	Sindrom Prader-Willi	Q87.14 Sindromul Prader-Willi	G01 MORFOMETRIE,G12 FOTOGRAFIERE,G25 MLPA ME028 PENTRU SINDROM PRADER-WILLI	2013-03-25	40106055507630372100	2019-01-09
739	Sindrom Prader-Willi	Q87.14 Sindromul Prader-Willi	G01 MORFOMETRIE	1994-05-24	40187099842673175983	2019-10-16
805	Scleroza tuberoasa	Q85.1 Scleroza tuberoasă	G01 MORFOMETRIE,G12 FOTOGRAFIERE	2010-04-13	40173527830537191273	2019-07-10
805	Scleroza tuberoasa	Q85.1 Scleroza tuberoasă	G01 MORFOMETRIE,G12 FOTOGRAFIERE	1974-05-25	40129456538482682304	2019-07-24
881	Sindrom Turner 45,X	Q96.0 Kariotip 45, X	G01 MORFOMETRIE,G12 FOTOGRAFIERE	2014-12-20	40841678887995061093	2019-08-06

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Cases

Detailed recommendations according to the Standard operating procedures and specification and implementation manual and an elaborate plan for extension to other centers in Romania were elaborated.

The number of registered cases per year has drastically increased since the beginning of the project:

At Month 12: 200 cases/year

At Month 30: 457 cases/year

At month 35: 580 cases/year

Future plans

Plan for extension to other centers in Romania

- identification of other centers ((Bihor, Bucharest, Cluj, Dolj) and hospitals with activity in rare diseases fields (C.I. Parhon Hospital Iasi – Nephrology)
- contact the representatives of centers/ hospitals
- preliminary discussion with representatives to present benefits of Orphacodes for rare diseases codification and periodic reports for Health Systems
- the representatives will indicate a team/ person involved in Orphacodes for rare diseases registration
- establish data for discussion with teams from other centers and major hospitals
- presentation of Orphacodes
- presentation of Standard operating procedures and specification and implementation manual
- presentation of IT tool (training)
- implementation of IT tool
- presentation of practical introduction of cases
- discussion about challenges
- establish monthly discussion about progress and difficulties
- statistical reports.

Final remarks

A simple, easy to use IT tool is essential for extensive ORPHAcodes implementation.