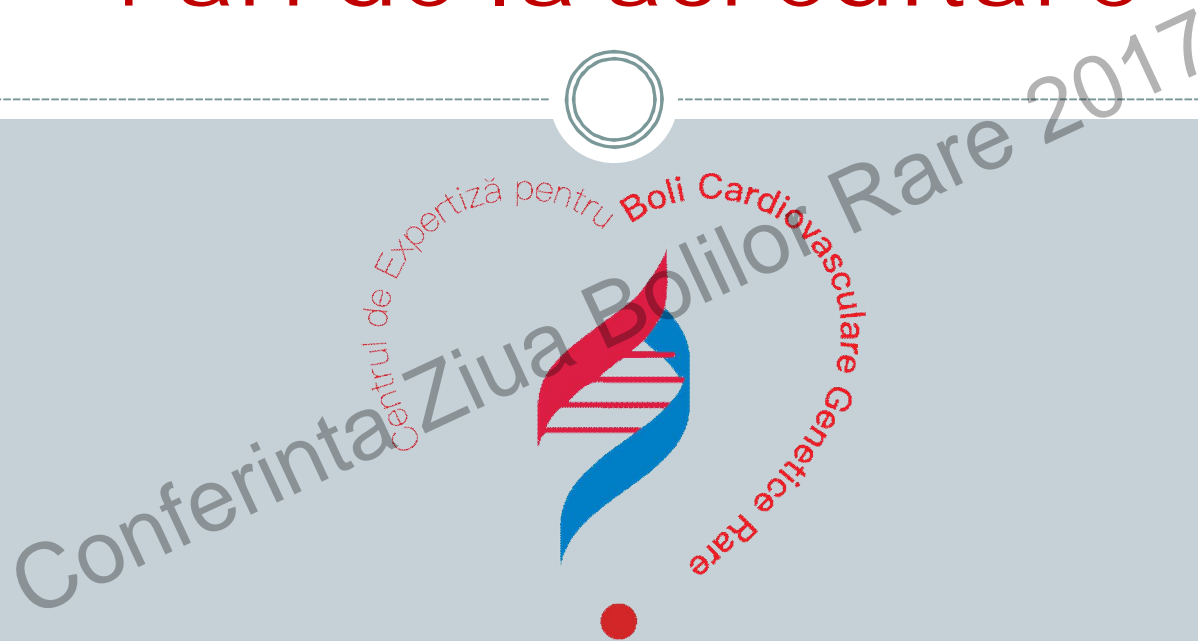


# Centrul de expertiza pentru boli cardiovasculare genetice rare - 1 an de la acreditare -



Conf.dr. Ruxandra JURCUT  
Coordonator

IUBCV "Prof.dr.C.C.Iliescu", UMF "Carol Davila"

# Bolile rare in cardiologie



- Definitia bolilor rare: prevalenta  $<1/2000$
- In cardiologie

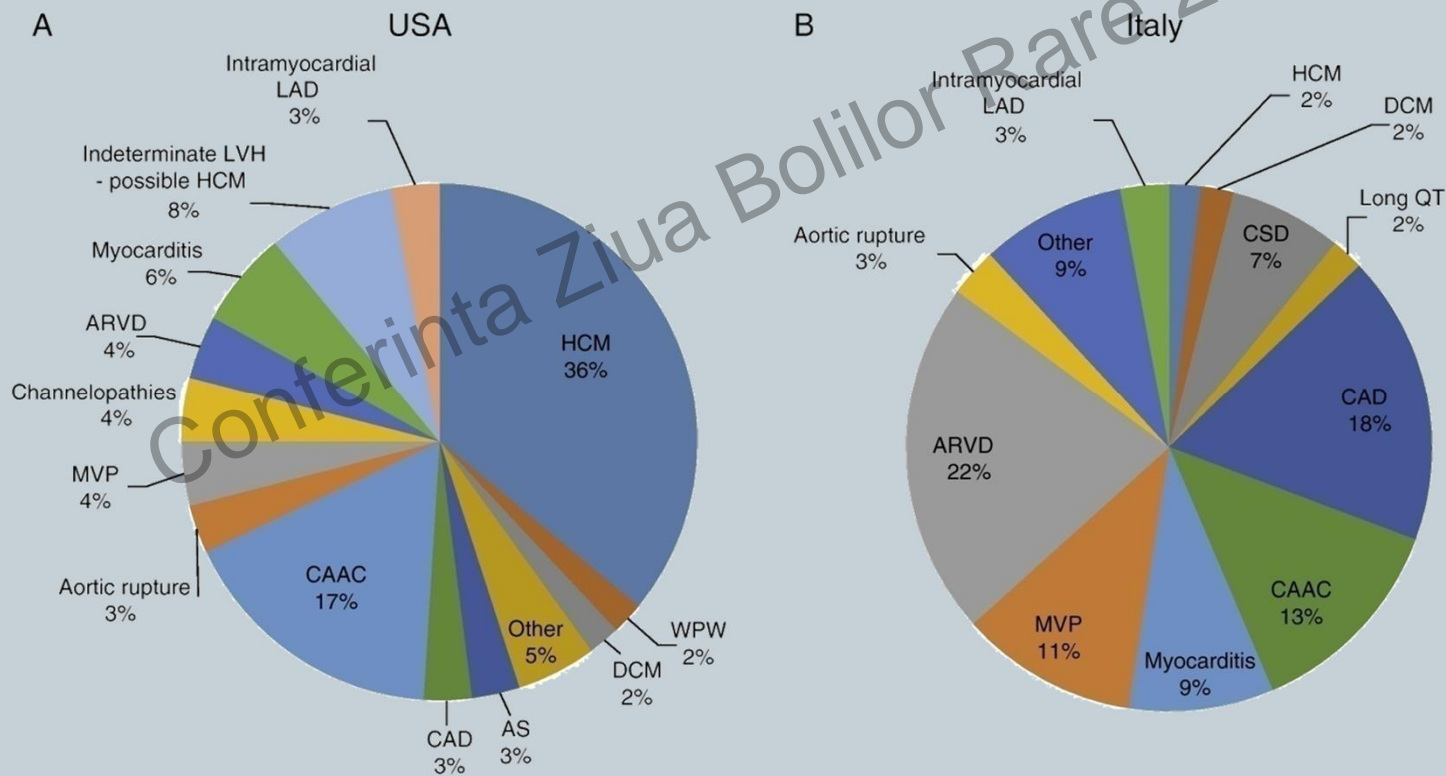
	Prevalenta
Displazia aritmogena de VD	1:2000-5000
Cardiomiopatia Fabry	1:17.000
Amiloidoza TTR	necunoscuta
Noncompactarea VS	1:10.000
Sindrom QT lung congenital	1:2500-5000
Sindrom Brugada	1:2000
CMH sarcomeric	1:500
Boala Marfan	1:20.000

Moarte subita  
Insuficienta cardiaca

Disectie de aorta

# Ce prevenim?

- Moartea subita
- Depistarea tardiva – crestere morbi-mortalitate



# CEBR acreditare nationala



MINISTERUL SĂNĂTĂȚII  
CABINET MINISTRU

Date: 15.06.2016

Country: ROMANIA

Institution issuing the endorsement letter: Ministry of Health

RE: Endorsement of Approved Healthcare Provider

This letter confirms that C.C. Iliescu Emergency Institute for cardiovascular diseases, Bucharest located at Sos. Fundeni Street, No 258, Sector 2, Bucharest, holding Sanitary Authorisation No. 771 of 20 May 2016, issued by Public Health Directorate of Bucharest, has been endorsed by Ministry of Health as an approved healthcare provider in Rare Genetic Cardiovascular Disorders in order to participate in European Reference Networks (ERN) for rare diseases, in accordance with the Romanian legal and regulatory requirements.

Sincerely,

MINISTER OF HEALTH  
VLAD VASILE VOICULESCU



Contact details:

Address: Intr. Cristian Popisteanu Street, no. 1-3, Bucharest

Email: [ministru@ms.ro](mailto:ministru@ms.ro)

Telephone: +40.21.307.26.90

MINISTERUL SĂNĂTĂȚII  
CABINET MINISTRU  
Nr. V.V.V. 697  
Ziua 15, Luna 06, Anul 2016

Conferința Națională de Polior Rare 2017

# Echipa



- Coordonator:
  - Conf.dr. Ruxandra Jurcut
- Membrii echipei:
  - Prof.dr. Carmen Ginghina
  - Prof .dr.Bogdan Popescu
  - Dr. Andreea Sandulescu
- Echipa extinsa
  - Genetica: Dr Viorica Radoi  
Centrul de Medicina Genomica UMF Timisoara
  - Psiholog IUBCV
  - Echipa cardiologie interventionala
  - Echipa electrofiziologie
  - Anatomie patologica

# Ce facem?



- Diagnostic complet
  - Inklusiv genetic, RM cardiac
- Screening familial
- Consiliere genetica
- Registre
- Retele europene
- Publicatii
- Cursuri si conferinte
- Educatie pentru pacienti



**NETWORK APPLICATION FORM**

5. List all the Healthcare Providers applying for membership in the Network Applicant and their location.

N°	Name of Healthcare Provider	Name of the Healthcare Provider Representative	Category of Facility, i.e. Regional Hospital, University Hospital, Private Clinic, etc.	Member State
1	Academic Medical Center	Arthur Wilde	University Hospital	Netherlands
2	Sant Joan de Déu - Barcelona Children's Hospital	Georgia Sarquella-Brugada	University Hospital	Spain
3	IRCCS Fondazione Salvatore Maugeri	Silvia Priori	University Hospital	Italy
4	Assistance Publique Hôpitaux de Paris	Philippe Charron	University Hospital	France
5	Hospital Universitario Puerta de Hierro Majadahonda	Pablo Garcia	University Hospital	Spain
6	CHU de Nantes	Vincent Probst	University Hospital	France
7	The Hospital District of Helsinki and Uusimaa	Jaana Pihkala	University Hospital	Finland
8	Motol University Hospital	Jan Janousek	University Hospital	Czech Republic
9	University Hospitals Leuven	Rik Wilems	University Hospital	Belgium
10	Ospedale Pediatrico Bambino Gesù	Fabrizio Drago	University Hospital	Italy
11	Umea University Hospital	Annika Rydberg	University Hospital	Sweden
12	Copenhagen University Hospital	Jacob Tfelt	University Hospital	Denmark
13	University Hospital Virgen Arrixaca	Juan Ramon Gimeno	University Hospital	Spain
14	University Hospital Munster	Eric Schulze-Bahr	University Hospital	Germany
15	St George's University Hospitals NHS Foundation Trust	Elijah Behr	University Hospital	United Kingdom
16	HCL hospital cardiologique CHU de Lyon	Philippe Chevalier	University Hospital	France
17	Emergency Institute for cardiovascular disease: Prof.dr.C.C.Iliescu	Ruxandra Jurcut	University Hospital	Romania
18	Azienda Ospedaliera di Rilievo Nazionale (AORN) dei colli - Ospedale Monaldi	Giuseppe Limongelli	University Hospital	Italy
19	Great Ormond Street Hospital, Foundation Trust	Juan Kaski	University Hospital	United Kingdom
20	Barts Heart Centre, St Bartholomew's Hospital	Perry Elliot	University Hospital	United Kingdom
21	IRCCS Istituto Auxologico Italiano	Peter Schwartz	University Hospital	Italy
22	IRCCS Foundation Policlinico San Matteo	Eloisa Arbustini	University Hospital	Italy
23	Universitair Ziekenhuis Brussel	Pedro Brugada	University Hospital	Belgium
24	SabUniversity of Padova - Azienda ospedaliera de Padovaino Illiceto	Sabino Illiceto	University Hospital	Italy

**Thematic area 1:**

Familial Electrical Diseases in Adults

- Long QT syndrome
- Brugada syndrome
- CPVT

**Thematic leader:**

Silvia Priori

**Thematic area 3:**

Genetic Cardiac Arrhythmias in Adults and Children

- Arrhythmic Cardiac Myopathy
- Hypertrophic Cardiomyopathy
- Dilated Cardiomyopathy

**Thematic leader:**

Philippe Charron

2017

# Echipa multidisciplinara boala Fabry



IUBCV   
"Prof.dr.C.C.Iliescu"

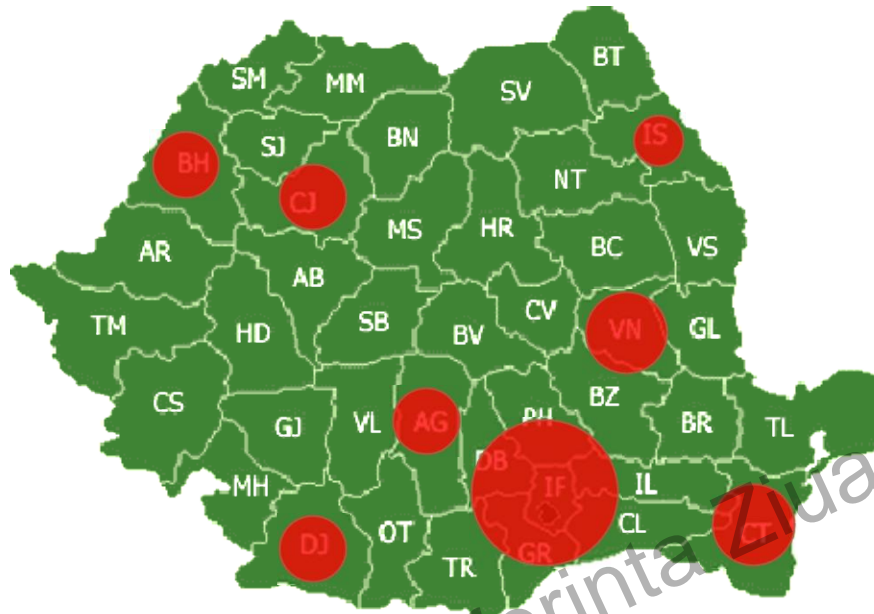


Cardiologie	Dr Ruxandra Jurcut
Nefrologie	Dr Elena Rusu, dr Camelia Achim, conf dr Gener Ismail
Neurologie	Dr Adriana Dulamea
Pediatrie	Dr Cristina Stoica
Oftalmologie	Dr Alice Munteanu

Conferinta Ziua Bolilor Rare 2017



# Expertiza boala Fabry in Romania



## CARDIOGEN Registrul National de Cardiomiopatii Genetice



CARDIOGEN reprezinta platforma nationala care va permite cunoasterea datelor de prevalenta si de standard de ingrijire pentru displazia aritmogena de ventricul drept, noncompactarea miocardica si cardiomiopatia Fabry.

CARDIOGEN si RENCMH au ca scop dezvoltarea investigatiilor de rezonanta magnetica cardiaca si testare genetica in cardiologie pe baza intelegerii magnitudinii problemei in Romania.

*Participarea dumneavoastra la acest registru este esentiala!*

[www.cardiogen.ro](http://www.cardiogen.ro)

Coordonator:  
S.L.dr.Ruxandra Jurcut, presedinte GL de Ecocardiografie si Alte Tehnici Imagistice

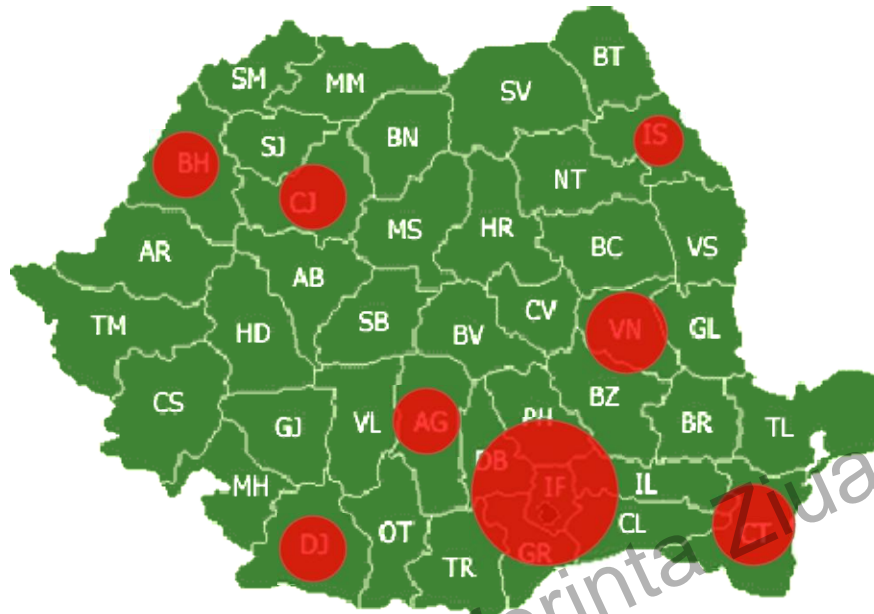
**FABRY REGISTRY Site Dashboard**

154099 - Jurcut RUCV

Filter by ID or Initials

Data Entry Activity (Past 90 Days)							
ID	Initials	Date	Details	ID	Initials	Date	Details
FR-206589	A-AI	14-Sep-2016	<a href="#">View</a>	FR-206587	S-S	14-Sep-2016	<a href="#">View</a>

# Expertiza boala Fabry in Romania



**CARDIOGEN** Registrul National de Cardiomiopatii Genetice

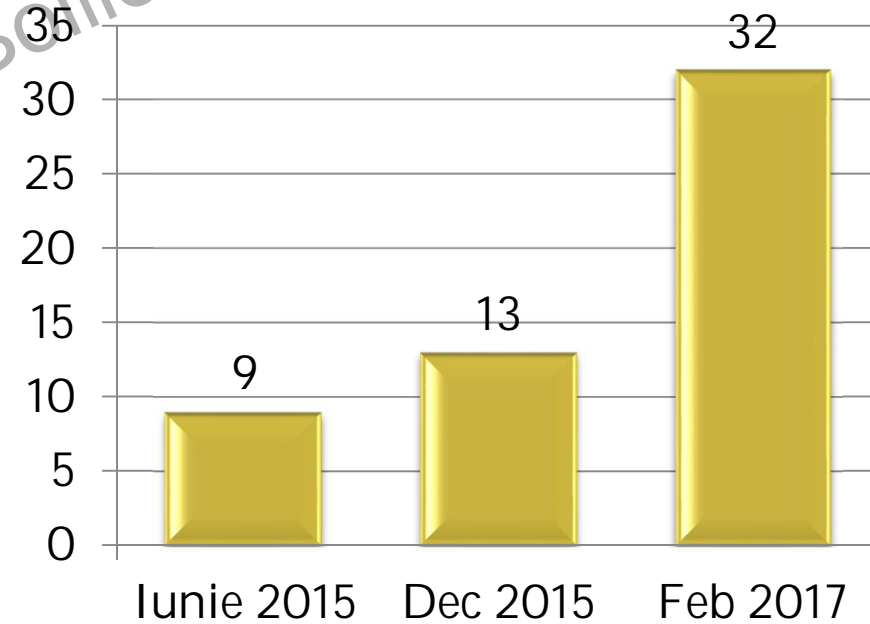
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Coordonator:  
S.L.dl. Ruxandra Jurcut, presedinta GL de Ecocardiografie si Alte Tehnici Imagistice



Nr	Patient	Sex	Age eval	Age diagn	Year Dgn	Diagnostic mode	Mutation
1	PDC	M	55	52	2013	Cardio HVS	p.Asn215Ser
2	PM	F	80	78	2014	Fam (cardio)	p.Asn215Ser
3	ID	M	43	37	2010	Renal	c.[671A>G](p.[Asn224Ser])
4	VM	F	26	26	2016	Family screening	c.[671A>G](p.[Asn224Ser])
5	SNP	M	40	36	2012	Renal	c.[141G>A], (p.[ W47*])
6	SS	F	59	59	2016	Fam (cardio)	c.[141G>A], (p.[ W47*])
7	MA	M	28	20	2008	Neuro (patie)	c.[1000_1290del]
8	MM	F	55	55	2016	Fam (cardio, neuro, ofta)	c.[1000_1290del]
9	MM	F	54	53	2015	Fam (?)	c.[485G>A] (p.[W162*])
10	MV	F	34	33	2015	Fam (Renal)	c.[485G>A] (p.[W162*])
11	MT	M	35	29	2010	Neuro	c.[485G>A] (p.[W162*])
12	BD	M	54	18	1978	Cutaneous (sud, ker)	c.[295C>T] (p.[Q99*])
13	BEle	F	31	31	2016	Fam (OK)	c.[295C>T] (p.[Q99*])
14	BEdu	M	2	2	2016	Fam (OK)	c.[295C>T] (p.[Q99*])
15	CI	M	46	36	2006	Neuro	c.[548delG]
16	CM	F	68	68	2015	Fam (cardio)	c.[548delG]
17	TI	F	48	48	2015	Fam (?)	c.[548delG]
18	LA	M	19	17	2013	Renal	c.[779G>A] (p.[G260E])
19	LM	F	45	40	2016	Fam (?)	c.[779G>A] (p.[G260E])
20	MIC	M	26	25	2015	Neuro	c.[836A>G](p.[Q279R])
21	MI	F	51	51	2016	Fam (neuro,ofta)	c.[836A>G](p.[Q279R])
22	EC	M	41	41	2016	Fam (Renal, ofta)	c.[836A>G](p.[Q279R])
23	PG	M	29	17	2004	Neuro	Unavailable
24	PI	F	59	47	2004	Fam (cardio, renal, ofta)	Unavailable
25	Jl	F	59	59	2016	Cardio	c.[644A>G](p.[N215S])
26	DC	M	41	41	2015	Renal	c.[863C>A] (p.[A288D])
27	DMR	M	49	49	2016	Renal	c.[831G>A](p.[Trp277*])
28	CM	F	78	70	2007	Fam (cardio)	Unavailable
29	BG	M	66	66	2016	Cardio	c.[416A>G](p.[Asn139Ser])
30	DV	F	48	48	2016	Neuro	

# Registre

- **Nationale**

- RENCMH **191 pts**

- CARDIOGEN

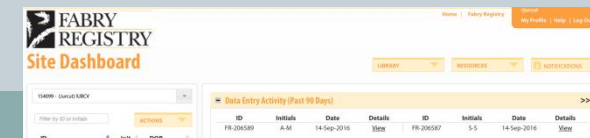
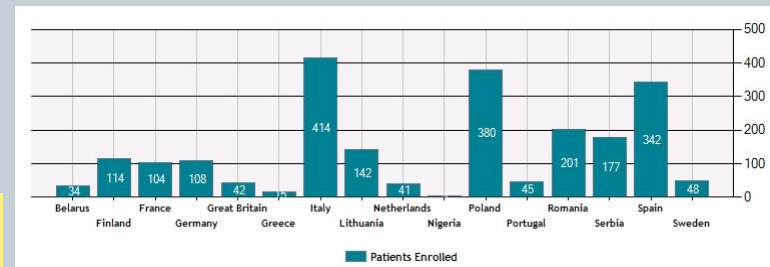
- ✦ Cardiomiopatie Fabry **13 pts**
- ✦ Displazie aritmogena VD **11 pts**
- ✦ Noncompactare cardiaca **4 pts**
- ✦ + boala Marfan



- **Internationale**

- EORP Cardiomiopatii **200 pts**

- Registrul international boala Fabry **9 pts**



# Educatie pentru medici



- Sesiuni dedicate bolilor CV genetice la
  - Congresul national de cardiologie
  - Congresul national de medicina interna
- Conferinte dedicatie bolilor CV genetice la
  - Cursurile postuniversitare ale Disciplinei Cardiologie IUBCV "Prof.dr.C.C. Diescu"
  - Cursurile pentru studenti
- Participare activa in expert meetings boala Fabry in Romania, Cehia

# Cercetare



- Studii asupra diagnosticului diferential al cardiomiopatiei Fabry de CMH sarcomerica
  - 1 lucrare doctorat
  - 1 lucrare de licenta (2017)
- Identificarea caracteristicilor cu determinism prognostic la rudele grd I ale pacientilor cu boala Marfan
  - 1 lucrare de licenta (2017)
- Evaluarea pacientilor cu ARVC – studiu de cohorta
  - 2 lucrari de licenta (2016, 2018)

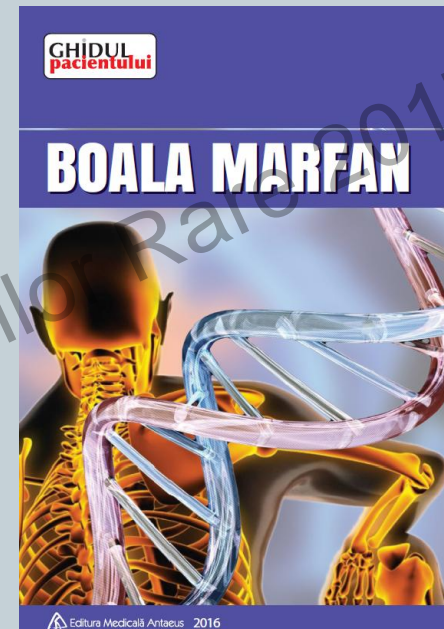
# Asociatii pentru pacienti



- Fundatia Romana pentru Boli Lizozomale
  - Asociatia pacientilor cu boala Fabry
- Comunitatea pacientilor cu CMH
  - Facebook

Conferinta Zilei Bolilor Rare 2017

# Ghiduri pentru pacienti



- Urmeaza
- Boala Fabry