



Help Lines  
for Rare  
Diseases

# EUROPEAN NETWORK OF RARE DISEASES HELP LINES

**-CALLER PROFILE ANALYSIS 2018-**

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## 1. Caller Profile Analysis October 2018

One of the goals of the European Network of Rare Diseases is to allow member help lines to exchange data across Europe. One of the criteria that members are asked to adhere to before becoming a member of the network is to record core call information in a uniform manner.

This makes it possible to compare data and exchange information amongst the network members. The initiative is particularly important considering the scarcity of available information on rare diseases.

As part of the network membership criteria, help lines are asked to meet certain criteria; one of these is to ensure that they are using the Orphanet coding system when recording call information.

This year the following 16 help lines took part in the exercise.

- Bulgaria - Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)
- Croatia – Croatian Help Line for Rare Diseases
- Denmark – Rare Diseases Denmark
- France - Maladies Rares Info Services (MRIS)
- France - AFM-Téléthon
- Hungary – Mentőöv
- Ireland - National Rare Diseases Office (NRDO)
- Italy - Università di Padova, Coordinating Centre for Rare Diseases, Veneto Region
- Italy – Telefono Verde Malattie Rare (national help line)
- Portugal – Linha Rara

- Romania - Romanian Myasthenia Gravis National Information Centre
- Romania - Romanian National Alliance for Rare Disease (NORO Help Line)
- Serbia – NORBS
- Spain - SIO Feder
- Switzerland - Info Maladies Rares (Lausanne)
- Switzerland – Seltene Krankheiten (Zurich)

Two new help lines are now part of the network: NORBS in Serbia, and Seltene Krankheiten (Zurich).

As help lines not only respond to calls but also to emails, we define as **enquirer** any person who calls, sends an email, writes a letter or visits the information service.

The following data were collected: the category of the enquirer, his/her age, his/her gender, the patient's age group, the type of contact, the reason for calling, the disease in question with its Orpha code, the response given, the time needed to respond, the region where the enquirer resides, a satisfaction score for the response given...

Each help line gathered the information over the month of October 2018. It is based on the analysis of a total of **1,700** calls, emails, letters or visits.

Enquiries were mostly about specific diseases. The enquiry discussed a specific disease in 1,185 calls or emails out of 1,700 or 70%. Of all enquiries, 177 were for undiagnosed patients (or 10%).

*Data were received from 16 help lines (Feder SIO, NORO Help Line, Linha Rara, ICRDOD, Romanian Myasthenia Gravis Info Centre, Coordinating Centre for Rare Diseases-Veneto Region, Telefono Verde Malattie Rare, Croatian Help Line for Rare Diseases, Maladies Rare Info Services and Info Maladies Rares, National Rare Diseases Office, Mentőöv, Rare Diseases Denmark, HelpLine Seltene Krankheiten, and NORBS):*

**Table 1: Participation of help lines in the Caller Profile Analysis since 2009**

Help Line	Country	2009	2011	2012	2013	2014	2015	2016	2017	2018	This year versus average (all other years)
		Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	Number of enquiries	
SIO-FEDER	Spain	239	163	203	255	303	293	316	205	366	+48%
CSH (Sjældne Handicap)	Denmark	72	-	17	-	-	-	-	-	-	-
Rare Diseases Denmark	Denmark								42	56	+28%
Maladies Rares Info Services	France	495	458	389	365	430	447	369	405	389	-7%
AFM-Téléthon	France	130	257	254	238	255	265	198	0	58	-71%
Uniamo	Italy	10	-	-	-	-	-	-	-	-	-
Linha Rara	Portugal	75	164	196	198	148	140	124	120	54	-63%
NORO	Romania	29	52	90	108	69	59	60	61	59	-11%
RadioOrg	Belgium	10	-	-	-	-	-	-	-	-	-
ICRDOD	Bulgaria	-	24	17	17	6	7	9	10	4	-69%
Myasthenia Gravis	Romania	-	25	28	28	29	25	24	26	27	2%
Info Maladies Rares	Switzerland	-	-	-	-	-	8	8	18	17	+50%
ENERCA	Spain/EU	-	3	3	2	5	4	5	-	-	-
Telefono Verde Malattie Rare	Italy	-	239	170	220	206	161	146	189	186	-2%
Croatian Helpline	Croatia	-	12	15	17	64	62	102	53	94	+102%

for Rare Diseases											
Coordinating Centre Veneto	Italy	-	342	294	224	331	161	226	395	289	+3%
NRDO	Ireland							5	8	12	+85%
Mentőöv	Hungary							32	41	42	+15%
NORBS	Serbia									36	NA
Seltene Krankheiten	Switzerland									12	NA
Total		1,060	1,739	1,676	1,672	1,850	1,714	1,633	1,573	1,700	+5%

Other help lines with no information are no longer member of the network or ceased their activities.

The overall number of enquiries is more or less stable since 2009 (+5%), however two dimensions need to be considered: the number of help lines participating in the Caller Profile Analysis (11 in 2011, 16 in 2018), and the number of enquiries received by each help line.

In 2011, help lines received an average of 165 enquiries; in 2018 the number of enquiries by help line dropped to 106 (-36% compared to 2011).

**Figure 1: location of the 16 help lines that participated in the Caller Profile Analysis 2018**



## 2. General information on the 16 help lines - CPA 2018

Table 2: characteristics of the help lines in 2018

Help Line		Creation year	Number of enquiries in 10/2018	Number of respondents	% fields completed	Overall satisfaction (highest 10)
AFM-Téléthon	France	2001	58	2	100	8.8
SIO-Feder	Spain	2001	366	10	100	8.3
ICRDOD	Bulgaria	2004	4	1	92.3	9.00
Linha Rara	Portugal	2008	54	3	92.3	NA
NORO help line	Romania	2008	59	2	100	8.8
Myasthenia Gravis	Romania	2006	27	2	100	9.3
Coordinating Centre Veneto	Italy	2002	289	6	100	9.2
Telefono Verde Malattie Rare	Italy	2008	186	3	100	7.7
Croatian help Line for rare Diseases	Croatia	2014	94	2	100	9.3
Maladies Rare Info Services	France	2001	389	3	84.6	NA
Info Maladies Rares	Switzerland	2015	17	1	100	9.2
NRDO	Ireland	2015	12	1	92.3	8.7
Mentőöv	Hungary	2015	42	2	100	9.0
Rare Diseases	Denmark	2016	56	5	92.3%	NA
NORBS	Serbia	2015	36	3	100	9.4
Seltene Krankheiten	Switzerland	2016	12	2	100	9.2
Total			1,700	48	97.1	9.0

The percentage of fields completed relates to the specific questionnaire used to collect the information. Overall, 97.1% of the information were provided by the helplines.

The Overall satisfaction is a score estimated by the respondent for each call. It corresponds to a self-evaluation of how the respondent handled the enquiry and is therefore subjective.

All together:

Help lines	Calls	Respondents
16	1,700	48
Number of enquiries by respondent (average)		35.4
Number of enquiries by help line (average)		106
Total time needed to respond (hours)		1,112

**Table 3: in October 2018, 16 help lines received 1,700 enquiries. 48 respondents replied, for an average of 35.4 replies/respondent. Large variability amongst help lines.**

Of note, all but one (Maladies Rare Infos Service) help lines are using the Orpha codes created by Orphanet/WHO for the classification of rare diseases. This is a major improvement since the first Caller Profile Analysis 2009-2010 where 4/8 help lines had implemented these codes.

	Orphacode
AFM Téléthon	Yes
SIO-Feder	Yes
ICRDOD	Yes
Linha Raras	Yes
Myasthenia Romania	Yes
NORO	Yes
Rare Diseases Denmark	Yes
Coordinating Centre Veneto Region	Yes
Telefono Verde Malattie Rare	Yes
Croatian Help Line	Yes
Maladies Rares Info Service	No
Info Maladies Rares	Yes
National Rare Diseases Office	Yes
Mentőöv	Yes
NORBS	Yes
Seltene Krankheiten	Yes

**Table 4: help lines that were using the Orpha Codes for rare diseases in October 2018**



### 3. Number of enquiries by population size

In total, the population served by the 16 help lines represented 251 million citizens. The 1,700 enquiries received represented 6.8 enquiries per million inhabitants, and this varied by country as shown in figure 2.

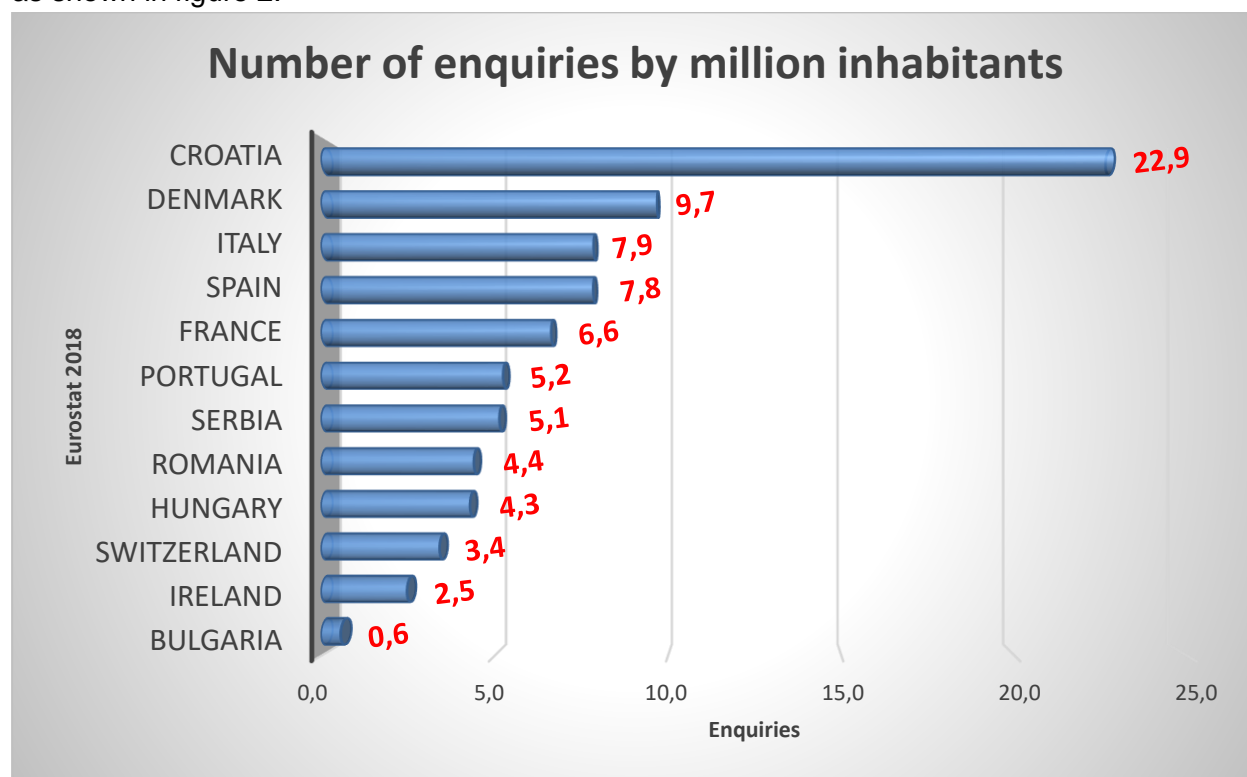


Figure 2: Number of enquiries by population size. Demographics are from Eurostat 2018. Median: 6

### 4. Age and gender

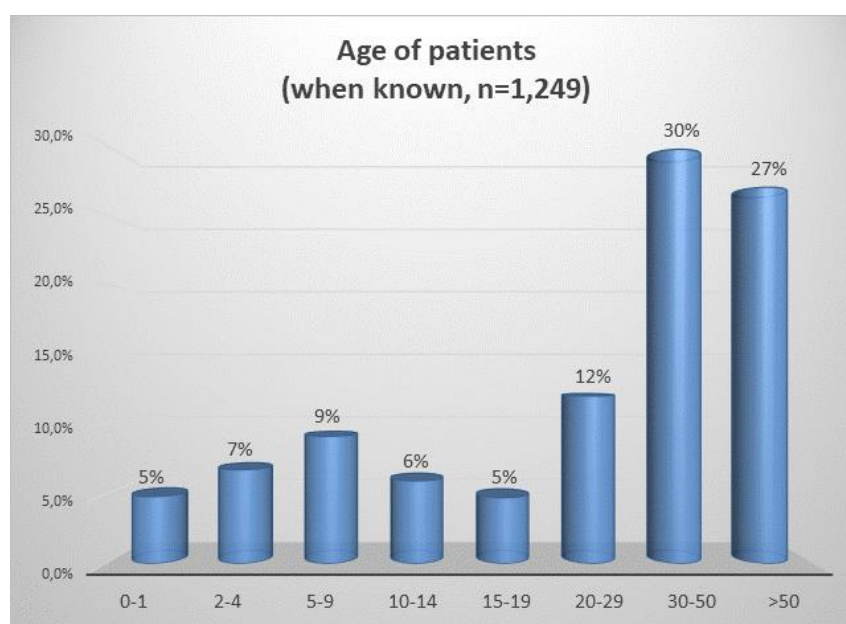


Figure 3: age of the patients in 2018 (1,249 data available)

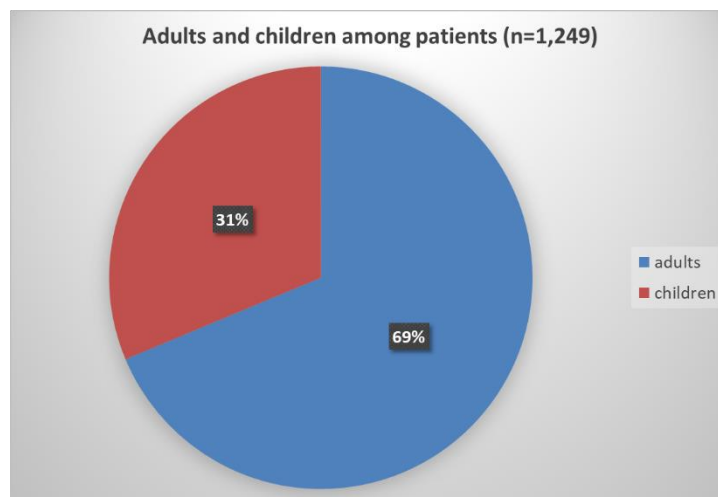


Figure 4: Adults and children among patients in 2018 (1,249 data available)

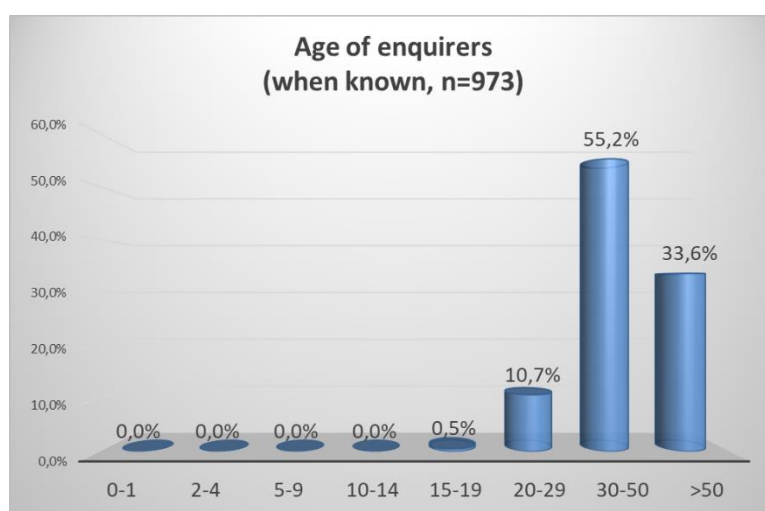


Figure 5: age of enquirers in 2018 (973 data available)

Most of enquirers were adults (10.7% were young adults, 55.2% from 30 to 50, and 33.6% more than 50).

As regards the enquirer's gender, the same distribution among men and women was observed in 2018 compared to previous years, with a larger number of enquiries made by women.

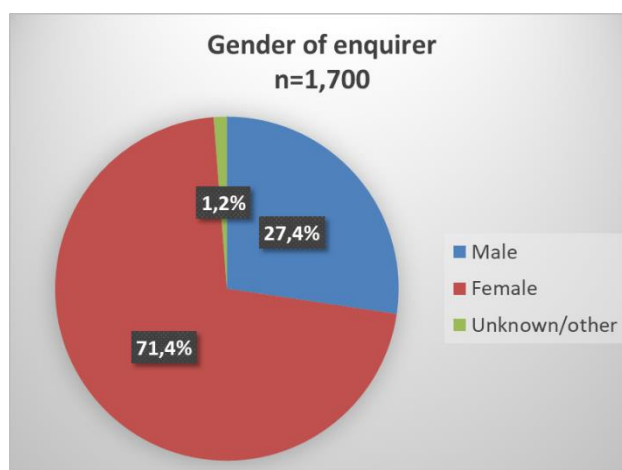
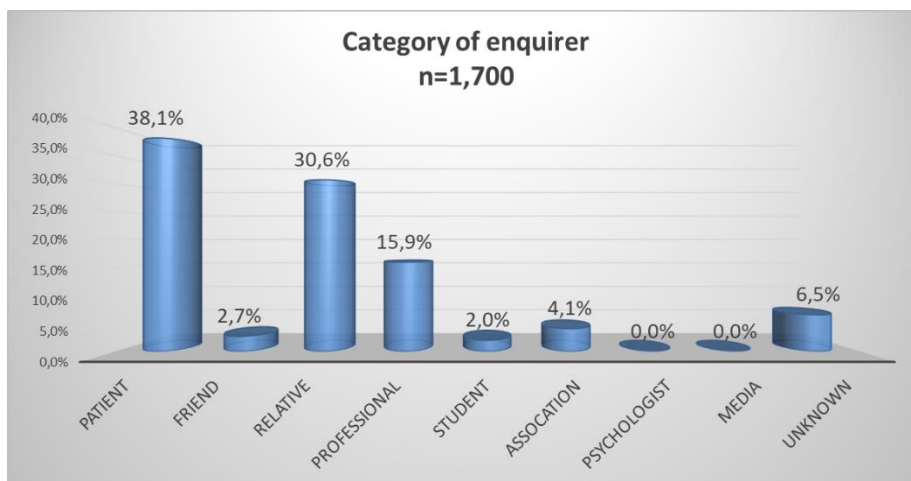


Figure 6: Men and women among enquirers in 2018

## 5. Category of enquirers

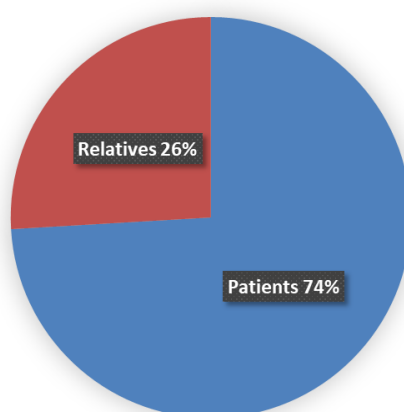
The enquirers did not differ in 2018 compared to previous years in terms of who they were.

Figure 7: category of enquirers in 2018.

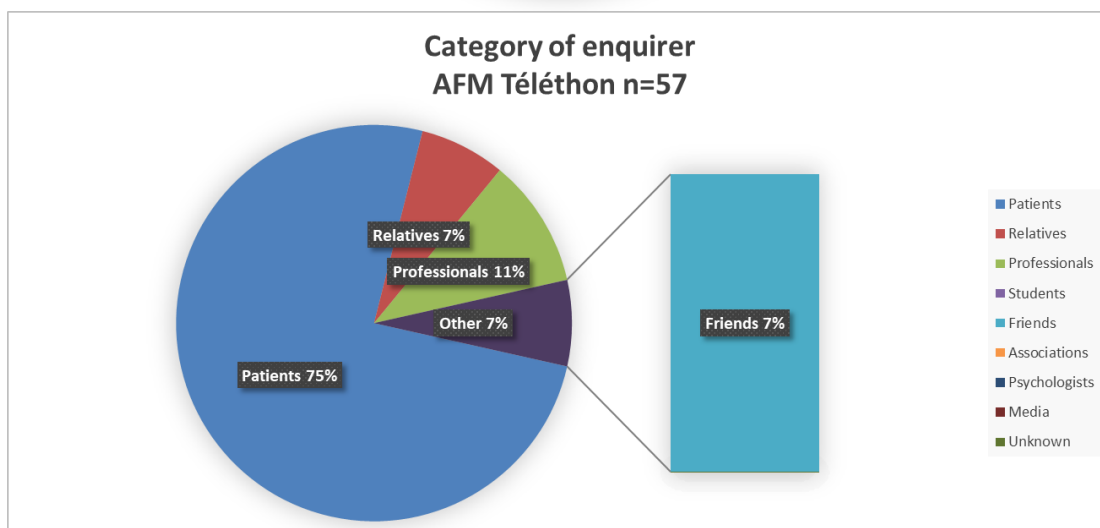


The diversity in help lines respective audiences is illustrated below: for each help line, the different types of enquirers are shown.

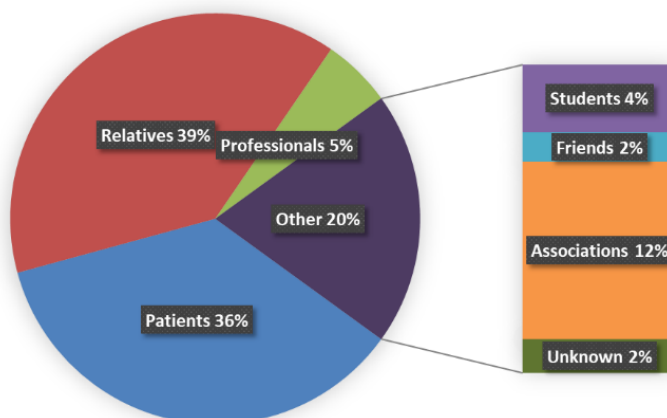
**Category of enquirer  
Myasthenia Gravis n=27**



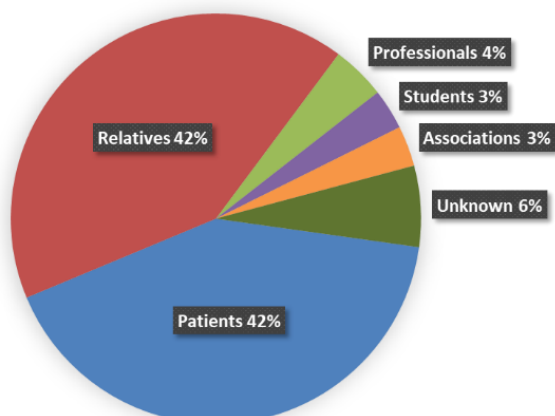
**Category of enquirer  
AFM Téléthon n=57**



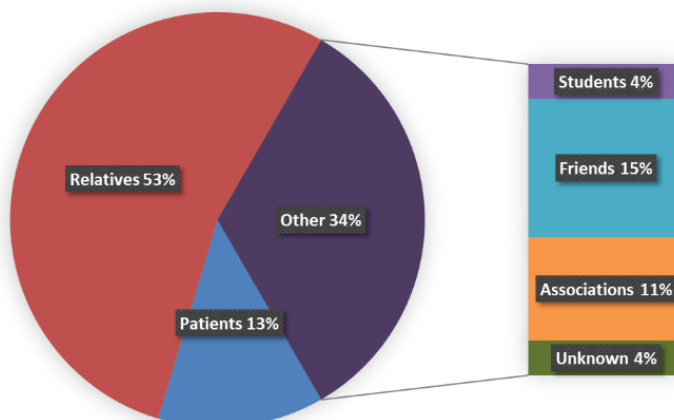
Category of enquirer  
SIO FEDER n=366



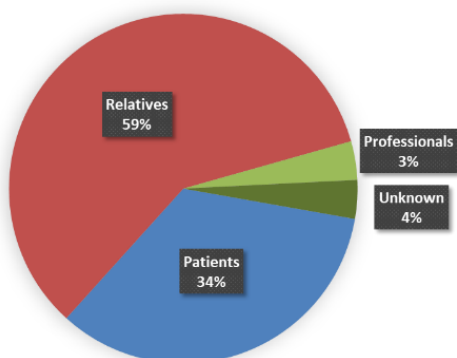
Category of enquirer  
Croatian HL n=94



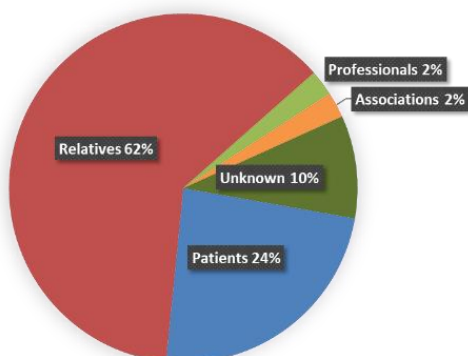
Category of enquirer  
Linha Rara n=54



Category of enquirer  
RDD Denmark n=56



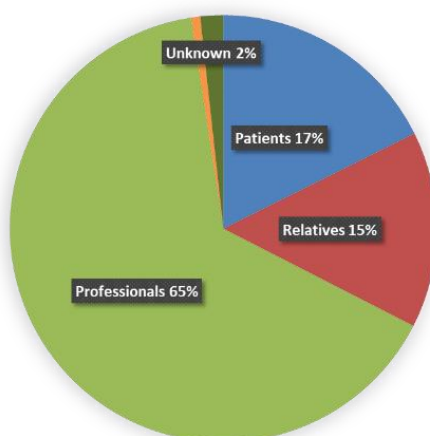
Category of enquirer  
Lifebelt n=42



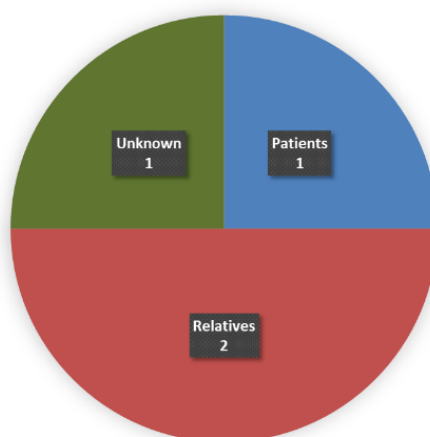
Category of enquirer  
National Rare Diseases Office n=12



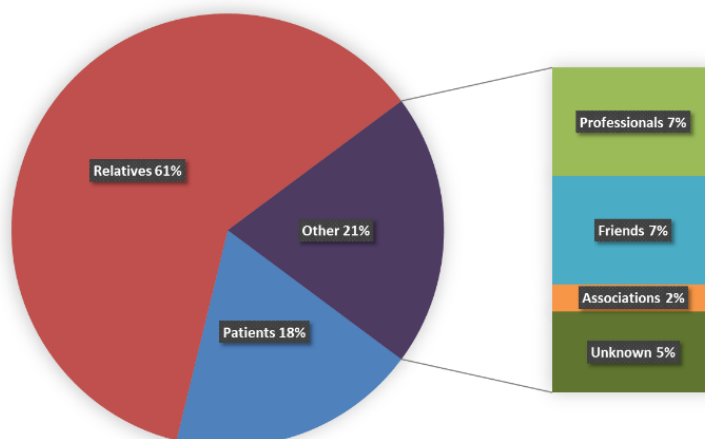
Category of enquirer  
Coordinating Centre Veneto Region n=289



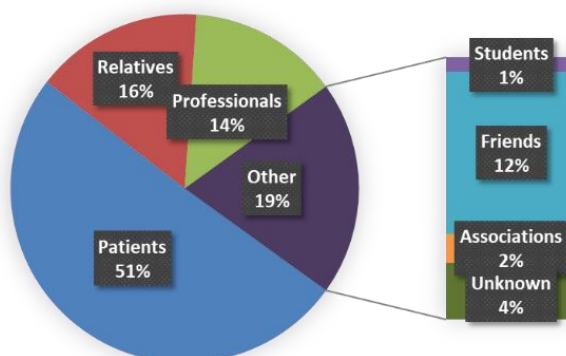
Category of enquirer  
ICRDOD n=4



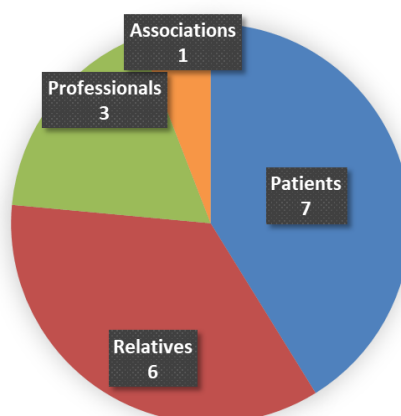
Category of enquirer  
NORO n=59



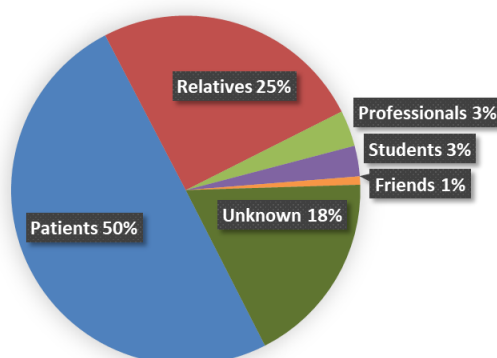
**Category of enquirer**  
**Telefono Verde Malattie Rare n=186**



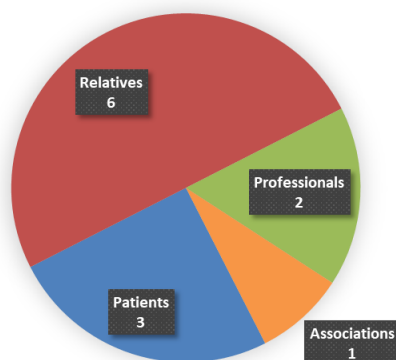
**Category of enquirer**  
**Info Maladies Rares Suisse n=17**



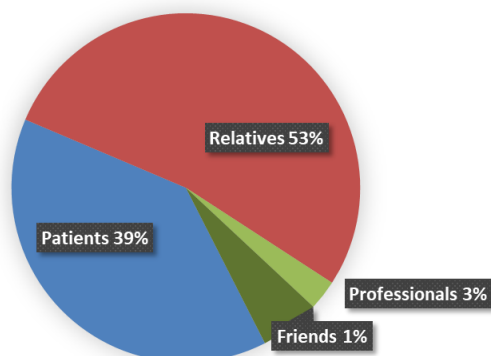
**Category of enquirer**  
**Maladies Rares Info Services n=389**



Category of enquirer  
Seltene Krankheiten n=12



Category of enquirer  
NORBS n=36

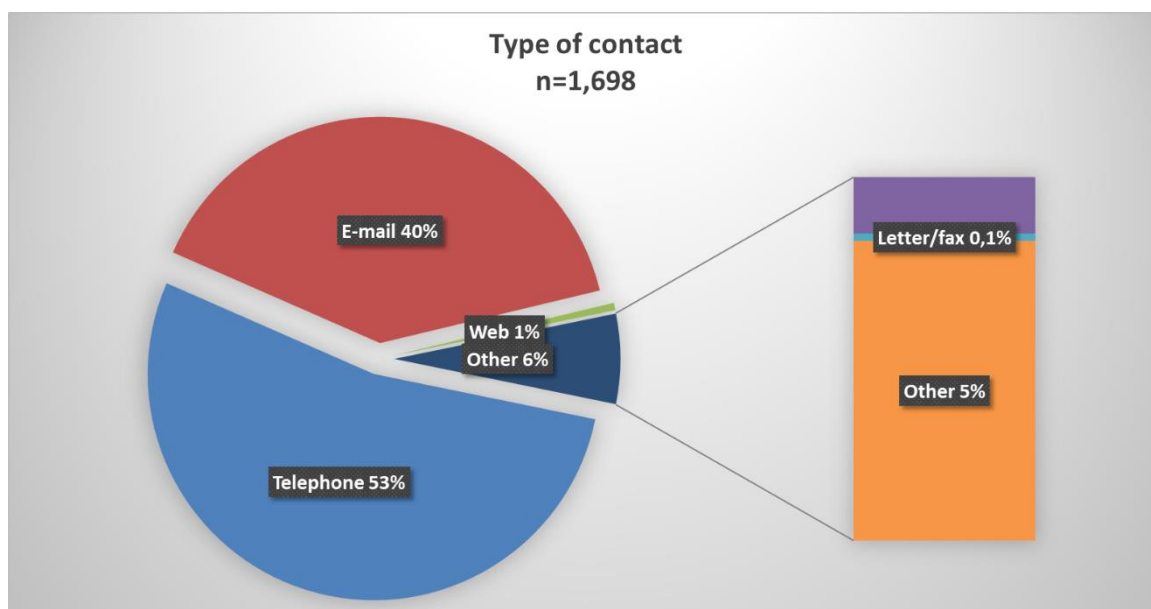




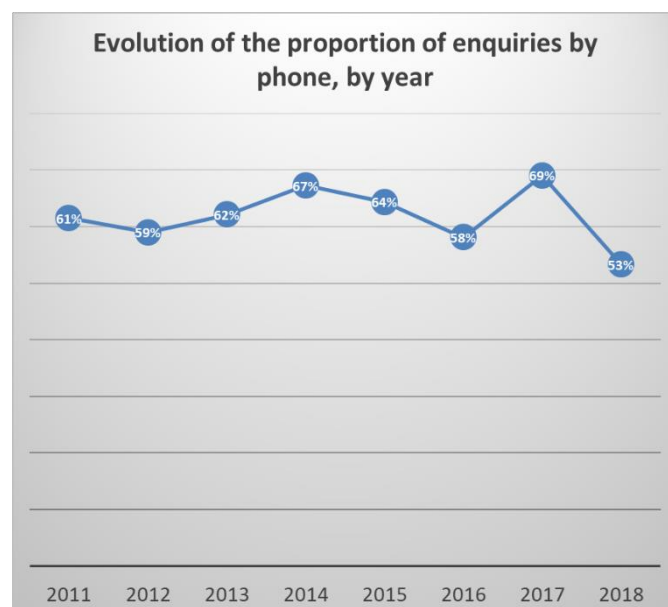
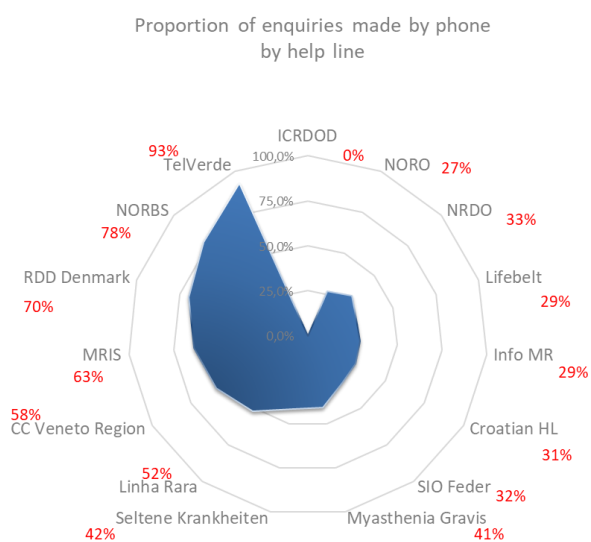
## 6. Type of contact

A majority of enquiries were made by telephone in 2018, as shown below:

**Figure 8: 53% of enquiries were made by telephone in 2018 (compared to 69% the previous year)**



**Figure 9: Some help lines operates almost exclusively by telephone (e.g. Telefono Verde Malattie Rare in Italy with 93% of enquiries by phone).**



In all age groups, telephone was the preferred mode for contacting the help line. Older age groups tend to call more often than to send emails. Younger enquirers used emails more often (51% e-mails users in the 15-29 age group, compared to 35% in the 30-50 and 35% in the >50).

### Type of contact by age group (n=966)

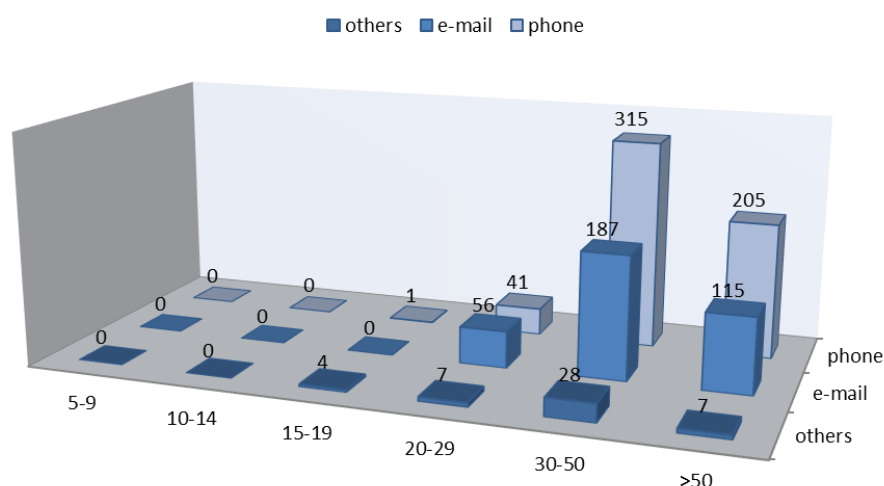


Figure 10: distribution of type of contact by age groups (966 data available)

## 7. Help Line awareness

In 2009, most of enquirers heard about the help line on internet (not shown). In 2018, enquirers heard about help lines firstly via other persons than healthcare professionals or patient organisations (friends, relatives... 28%), secondly via healthcare professionals (23%), the media (20%), patient organisations (14%), internet (11%). Among professionals, other health care professionals are the main source of information on the existence of the help line (for 83%, figure 12).

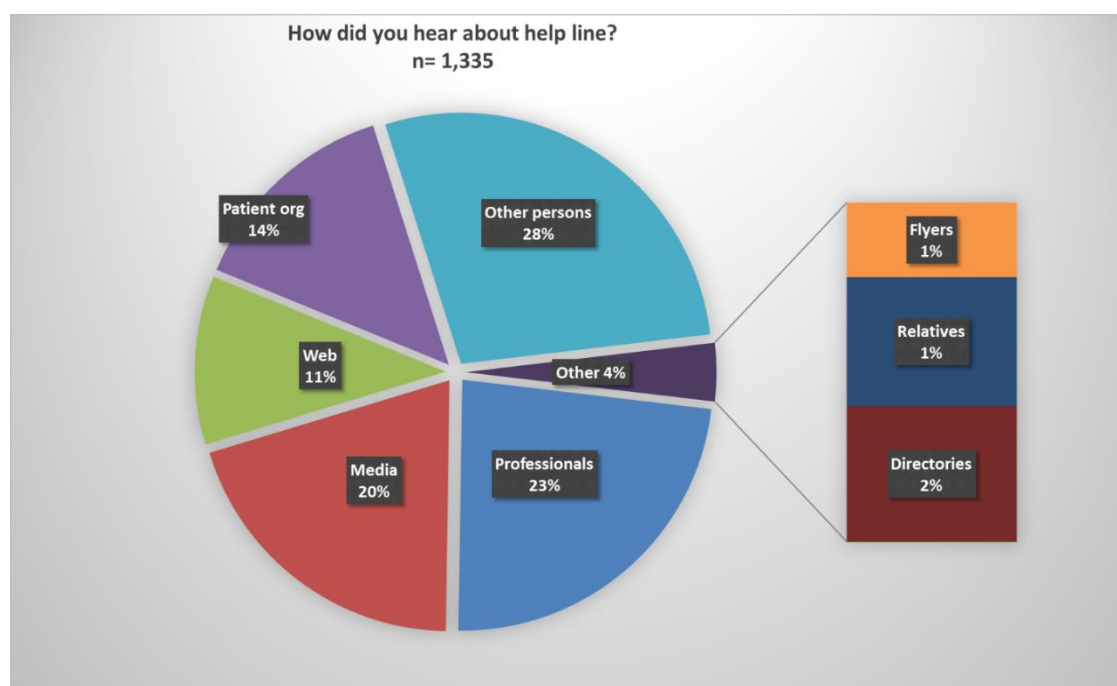
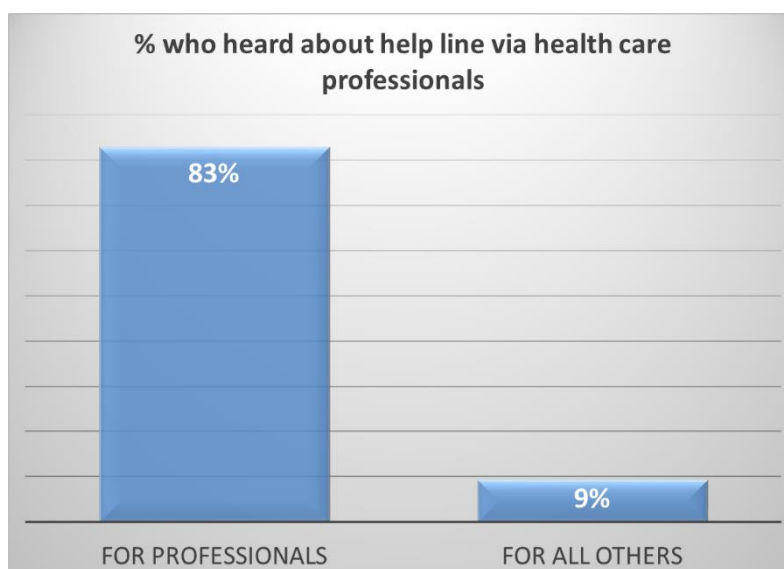
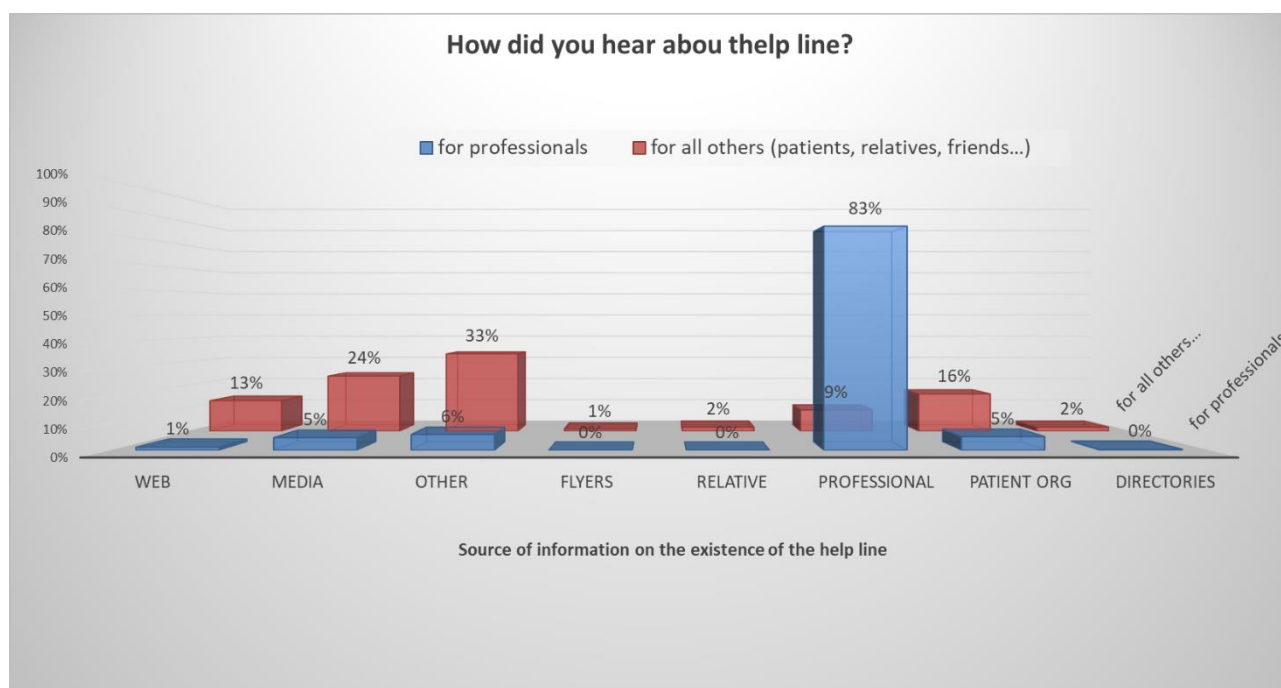


Figure 11: 11% of enquirers were informed about the help line via internet versus 28% by friends or relatives, 20% by the media, 23% by healthcare professionals, 14% by patient organisations in 2018



**Figure 12: proportion of enquirers who heard about the help line via health care professionals. While 83% of healthcare professionals are informed of the service by other healthcare professionals, other enquirers were rarely informed by health care professionals (9%)**

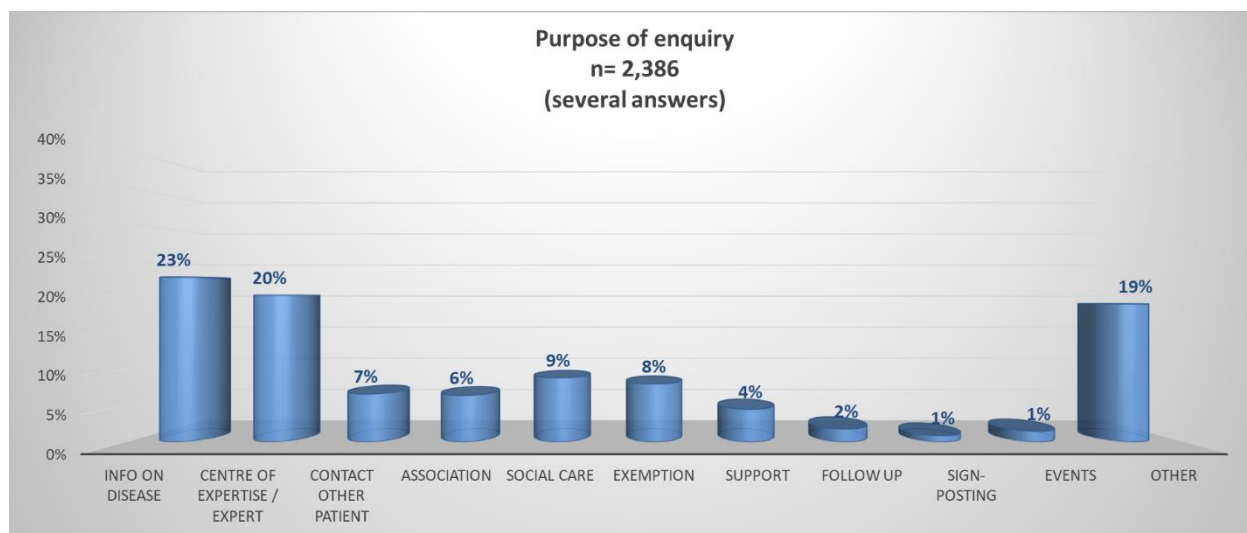


**Figure 13: how did you hear about the help line? Distribution of sources of information for health care professionals who contacted the help line versus other.**

## 8. Purpose of enquiry and response given

The purpose for contacting a help line was not significantly different in 2018 from previous years. Request for information on the disease remained the main purpose, and information on centres of expertise and/or experts was the second reason for contacting a help line (several reasons can lead to contact a help line, reason why the number of reasons exceeds the number of enquiries).

Figure 14: purpose of enquiry in 2018



Similarly, different responses could be given to the enquirer, and 3,265 information/responses were given to 1,700 enquiries:

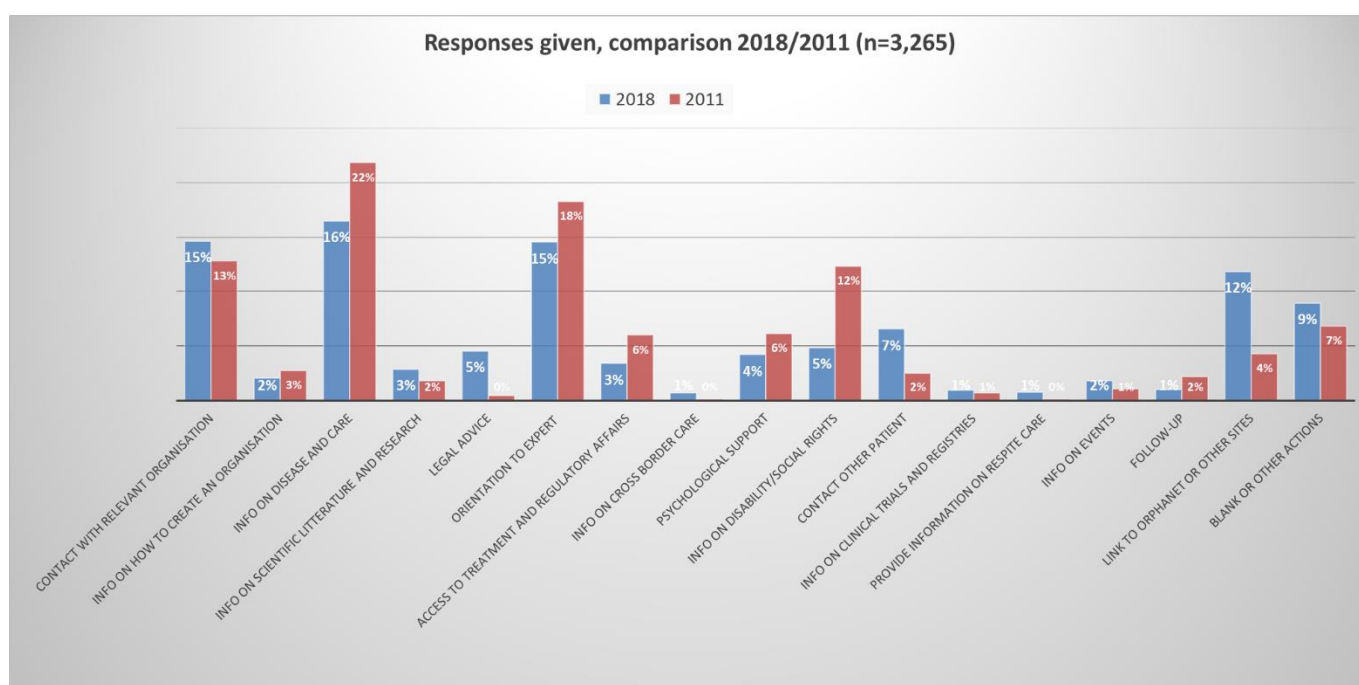


Figure 15: responses given in 2018 compared to 2011

## 9. Duration of the enquiry

The information on the duration of the query was available for 1,699 calls, emails, letters, or visits. In average, queries lasted for 39 minutes (SD 81.4) and median was 23. Note that the distribution of duration is not normal:

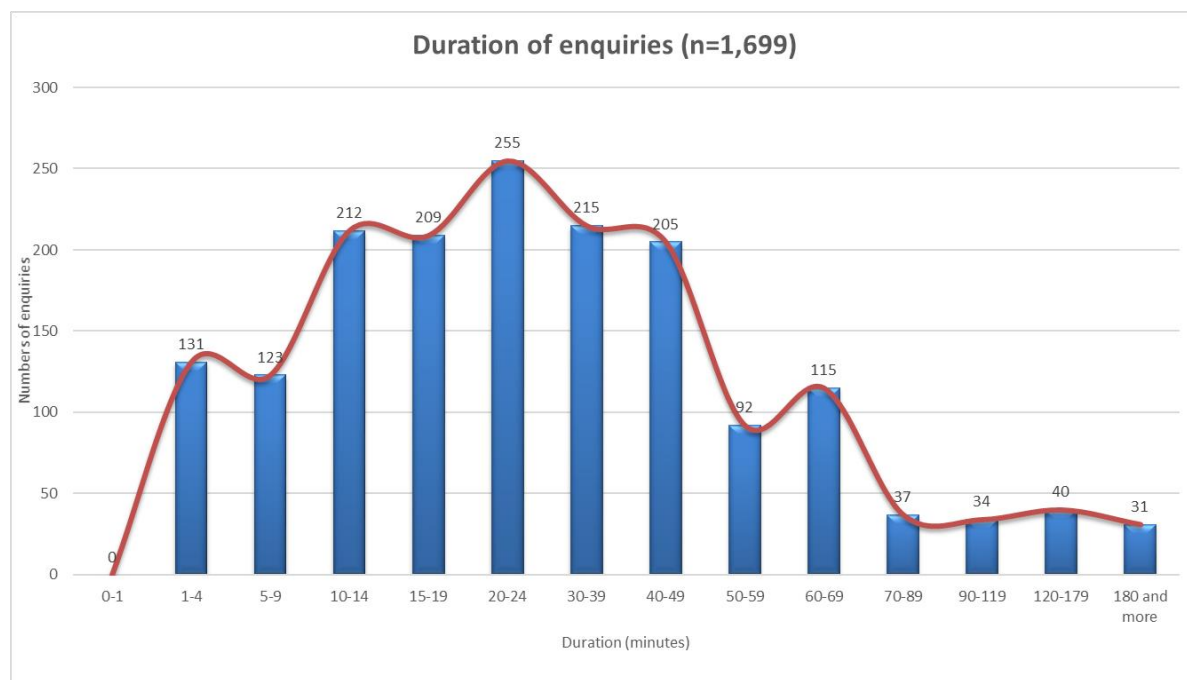


Figure 16: distribution of the duration of queries in 2018

Even though 50% of the calls lasted for 23 minutes or less, the proportion of calls or emails that needed more than 40 minutes was high (31%); 60 minutes or more represented 15% of enquiries.

For help lines who could collect that information, we could estimate the average duration according to the type of contact used, as shown below:

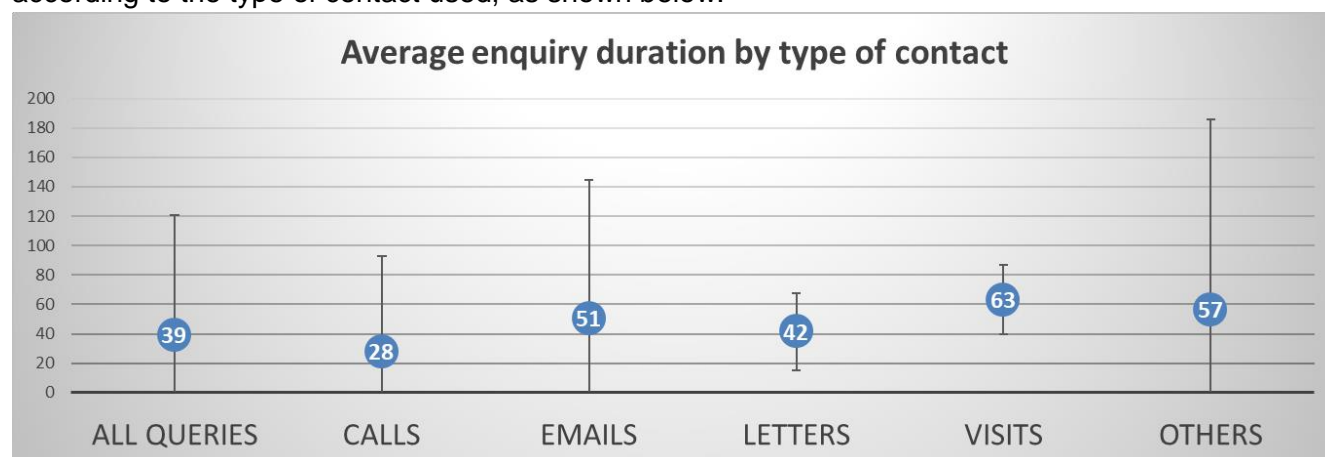


Figure 17: mean duration of calls, emails or others responses, by type of contact, in 2018.

These estimates were calculated from 906 calls, 674 emails, 17 visits, and 72 others for which duration was indicated. For the duration of calls, this did not include the time needed to prepare additional responses sent by email or by mail.

## Duration by help line

We can estimate the average and median duration of each enquiry (shown below). The duration of an enquiry varied greatly within help lines, even if the average is 39 minutes, some calls or emails may require several hours to respond to. One help line is not shown: Seltene Krankheiten, with a median duration of 240 minutes.

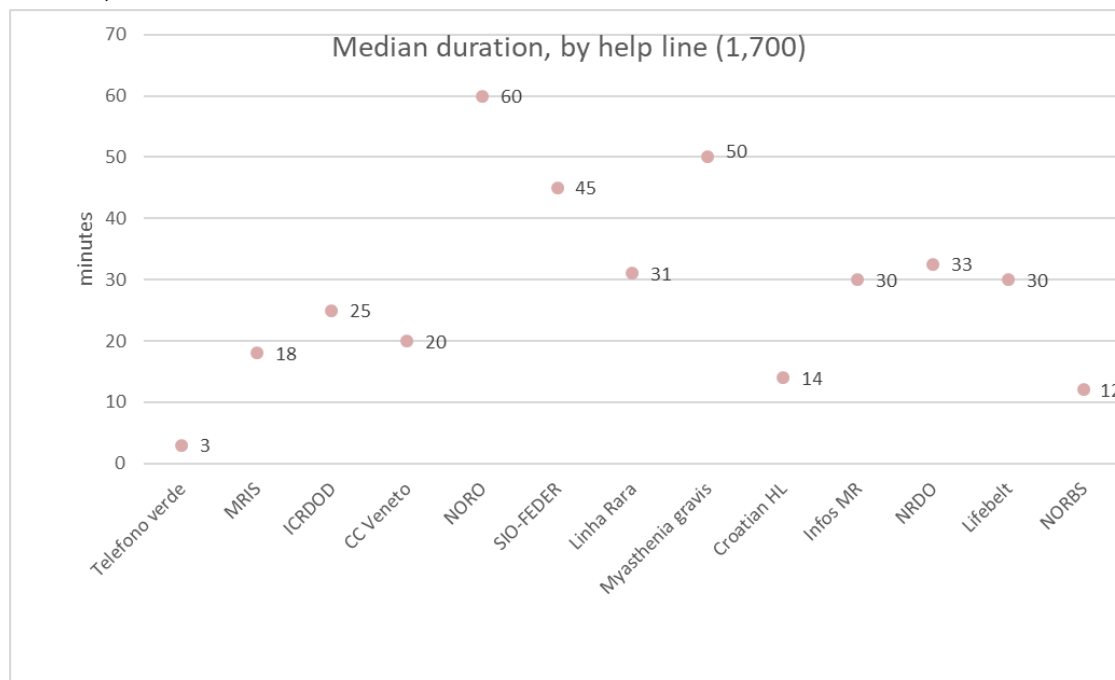


Figure 18: the median duration of an enquiry varied by help line

## 10. Self-evaluation of the quality of the response given by the respondents

Respondents were invited to score the quality of the call/email on a scale from 0 (could not have been worst) to 10 (could not have been better).

The distribution of the scores is shown below:

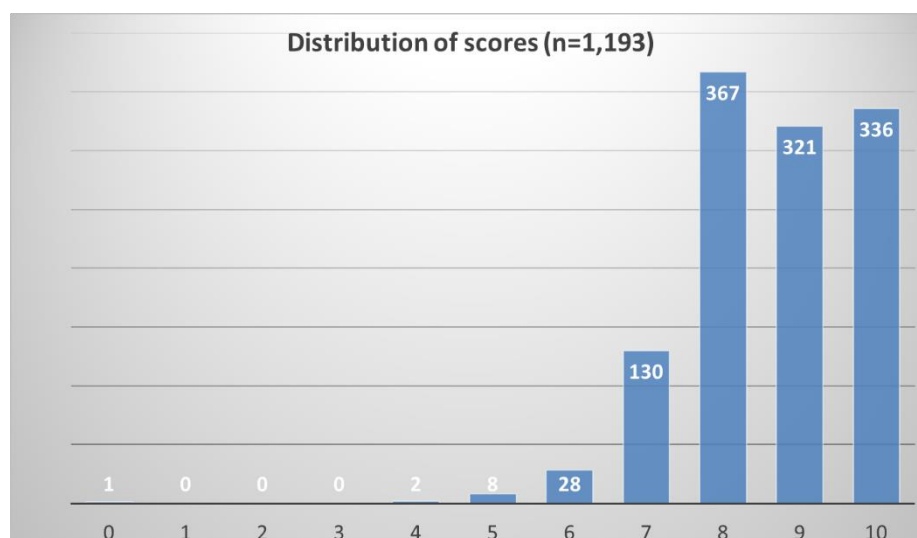
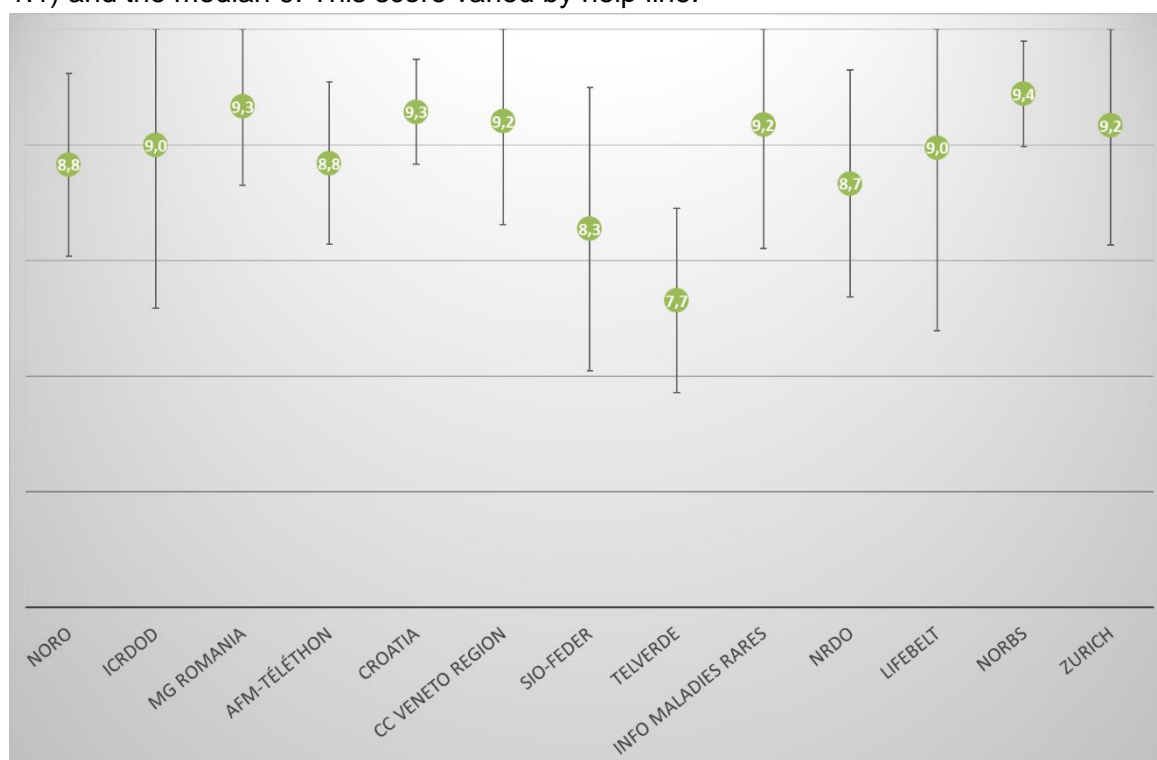


Figure 19: distribution of scores by respondents, from 0 to 10.

Not all help lines ask their respondents to grade the quality of the interaction and response given, reason why the total equals 1,193 scores for 13 lines (Linha Rara, Maladies Rares Info Services and Rare Diseases Denmark did not collect this data. The mean score was 8.7 (SD: 1.1) and the median 9. This score varied by help line:



**Figure 20: average score (SD) by help line. Satisfaction measures to which extend respondents are satisfied with the information they gave or the way they led the discussion, based on a scale from „0“ (very unsatisfied) to „10“ (very satisfied. Tilverde is for Telefono Verde Malattie Rare, Italy)**

## Acknowledgements

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- Rare Diseases Coordinating Centre Veneto Region: Dr Monica Mazzucato, and Prof Paola Facchin
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- NORO Help Line: Dorica Dan, Zsuzsa Lazar, Florina Breban
- Croatian Help Line for Rare Diseases: Vlasta Zamek, Kristina Maurer
- Myasthenia Gravis Romania: Nadia Radulescu
- Linha Rara: Rute Cardoso, Maria João Freitas
- ICRDOD: Georgi Iskrov, Samanta Georgieva
- SIO Feder: Estrella Mayoral Rivero, Isabel Fernández Aldeguer
- Maladies Rares Info Services: Thomas Heuyer
- Info Maladies Rares: Loredana D'Amato Sizonenko, Alessandra Strom
- AFM-Téléthon: Tuy Nga Brignol, Marguerite Friconneau, Annie Archer
- NRDO Ireland: Jacqueline Turner
- Mentööv: Gabor Pogany
- Rare Diseases Denmark Help Line: Lene Jensen, Stephanie Jøker Nielsen
- NORBS: Andrea Nenadic

- Help Line Seltene Krankheiten Zurich: Sabrina Strebel

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European Commission, Public Health Programme,  
DG Health and Consumer Protection  
Programme of Community action in the field of public health



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