



CENTRO SERVIZI MALATTIE RARE

Rare diseases help desk



ENSA General Assembly 2024
Brussels 22, 23 and 24 January 2024



sportello

Lunedì e Giovedì
h. 09.00 - 14.00

riceve anche su appuntamento

tel: **+39 376 2113589**

info:centroservizimalattierare@gmail.com

www.csmr-centroservizimalattierare.eu

Dott.ssa Roberta Bresciani
Vice-president association p63 syndrome
E.E.C. International
Padova – Veneto - Italy

A few examples of requests...

Good morning,

I am the partner of a 28-year-old girl living in Padua who started a few years ago on the path to diagnosis for a suspected rare disease, a process which is still ongoing. Since her situation is invalidating due to the continuous pain she has to endure, **we have sought psychological support**, but the professionals we have turned to, who are not experts in the complex sphere of rare diseases, have not been able to give her the relief we hoped for. **Having found your contact details on the internet I am turning to your service for information and advice.**

I am writing to you because I have been sick for 13 years and 13 years of blindly consulting doctors without finding a solution. **I practically cannot get a diagnosis and therefore cannot get a cure. Could you help me?**

Perhaps point me to a specialist or facility of yours where I can explain my situation in the hope of getting to the bottom of it?

Thanking you,
a cordial greeting.

A few examples of requests...

Good morning,

I am writing to you on behalf of my wife, asking for a listening appointment to improve or solve her pathology. Our idea is to see if there is anyone who has suffered from a similar problem and has solved or even improved it. If you could help us with a specific address, we thank you in advance.

Dear,

I was given your contact details in the Municipality as I have been diagnosed with Ehlers-Danlos Syndrome in 2020.

I am turning to you to see if you can help me understand how to move forward as I am at a standstill both medically and at work.

Thank you for all the help you can offer me.



What it is:

There is a place where children with rare diseases become children again.

A concrete point of reference for people with rare diseases, enabling the basic principles of health and supporting social emergencies thanks to experienced operators.

Our targets:

Improving the **quality of life** of people affected by rare diseases.

Contributing to **information, networking and protection of the rights** of rare disease patients, in health, social and socio-health policies, for the protection of people with disabilities, victims of discrimination.

Facilitating **the access of patients with RD to all the benefits** provided by law in this field. Facilitating access of RD patients to treatment

Spreading the culture on rare diseases, encouraging specialists to apply the protocols shared by inter-regional reference centres.

Actively cooperate with the Public Relations Office (OPR) in setting up a permanent discussion table with the main associations of patients with RD

Creating information, and training courses for individuals for families and operators, collaborating with a national and international network.

Informare i pazienti sulle novità farmaceutiche sperimentali ove possibile, solo in relazione a collaborazioni certificate

Manage operational activities to facilitate social integration, maintaining constant collaborative contacts also with the Regional Coordination for Rare Diseases of Veneto and the Ministry of Health.

What it is:

Primary services

First reception and/or counselling both in physical presence and online, according to patients' requests.

Counselling service

Taking charge of the subject, thanks to certified specialists and centres of expertise.

Health-social support and integration through inclusion in new generation activities and projects.

The activity is carried out from Italy, collaborating with specialists throughout Europe and beyond.

CONTACTS:

Mail: centroservizimalattierare@gmail.com

24-hour telephone: +39 376 2113589

What it is: the person at the centre



Why:

- Rare diseases, a common reality in today's society, a real social and health emergency that affects more than 38.000 people in the Veneto region and almost 2 million people in Italy.
- There are 7.000-8.000 recognised rare diseases and as many unrecognised pathologies. Loneliness, misinformation, misdiagnosis, wrong drugs, lack of treatment plans and care, discrimination, social estrangement are just some of the issues we address with our associates.
- Since 2018, they have requested the opening of the help desk:

Belluno (Feltre e Pieve di Cadore)

Venezia (Chioggia)

Vicenza(Bassano, Cittadella,
Thiene...)

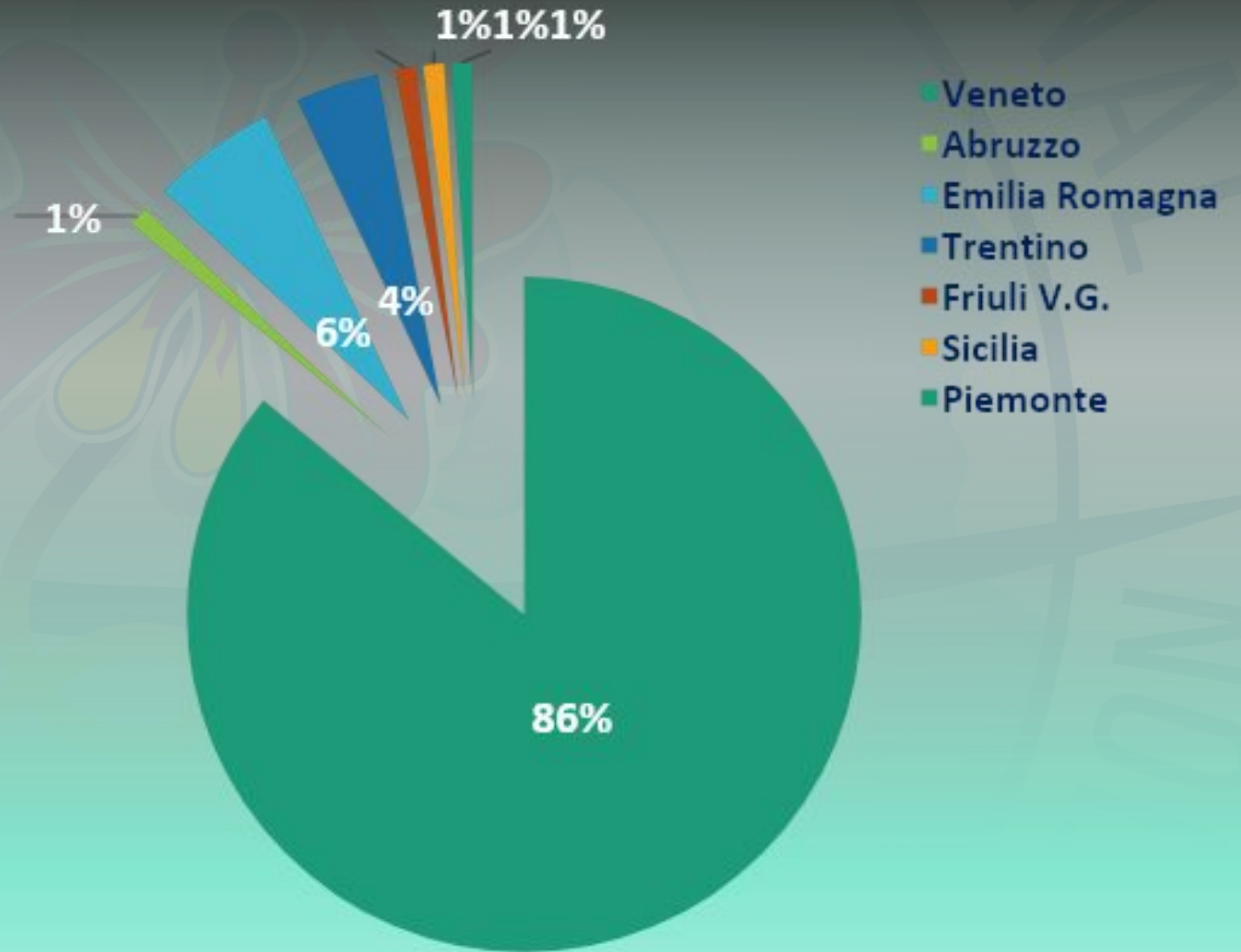
And many more

User graphs

CSMR

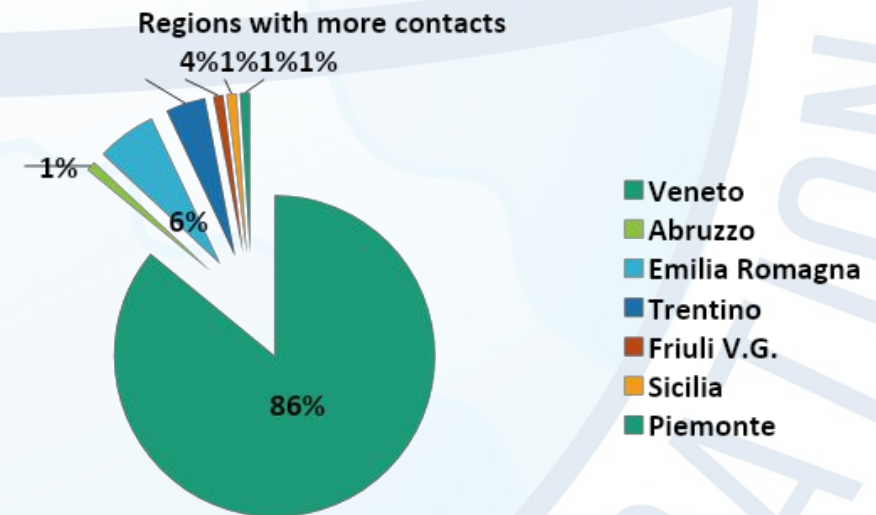
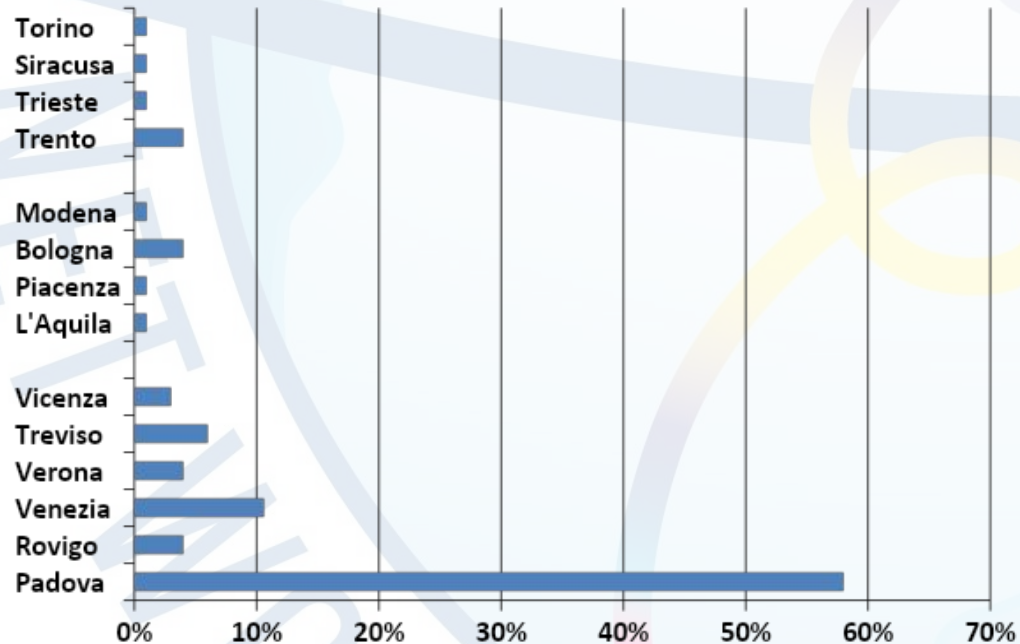
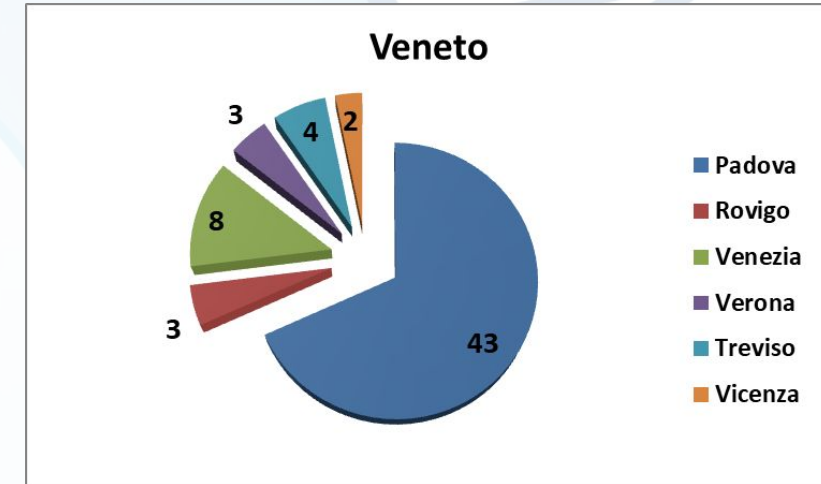
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Regions with more contacts



Numbers (Reference year 2022)

- Over 400 telephone contacts by e-mail;
- More than 200 physical contacts at the counter;
- 100 intakes

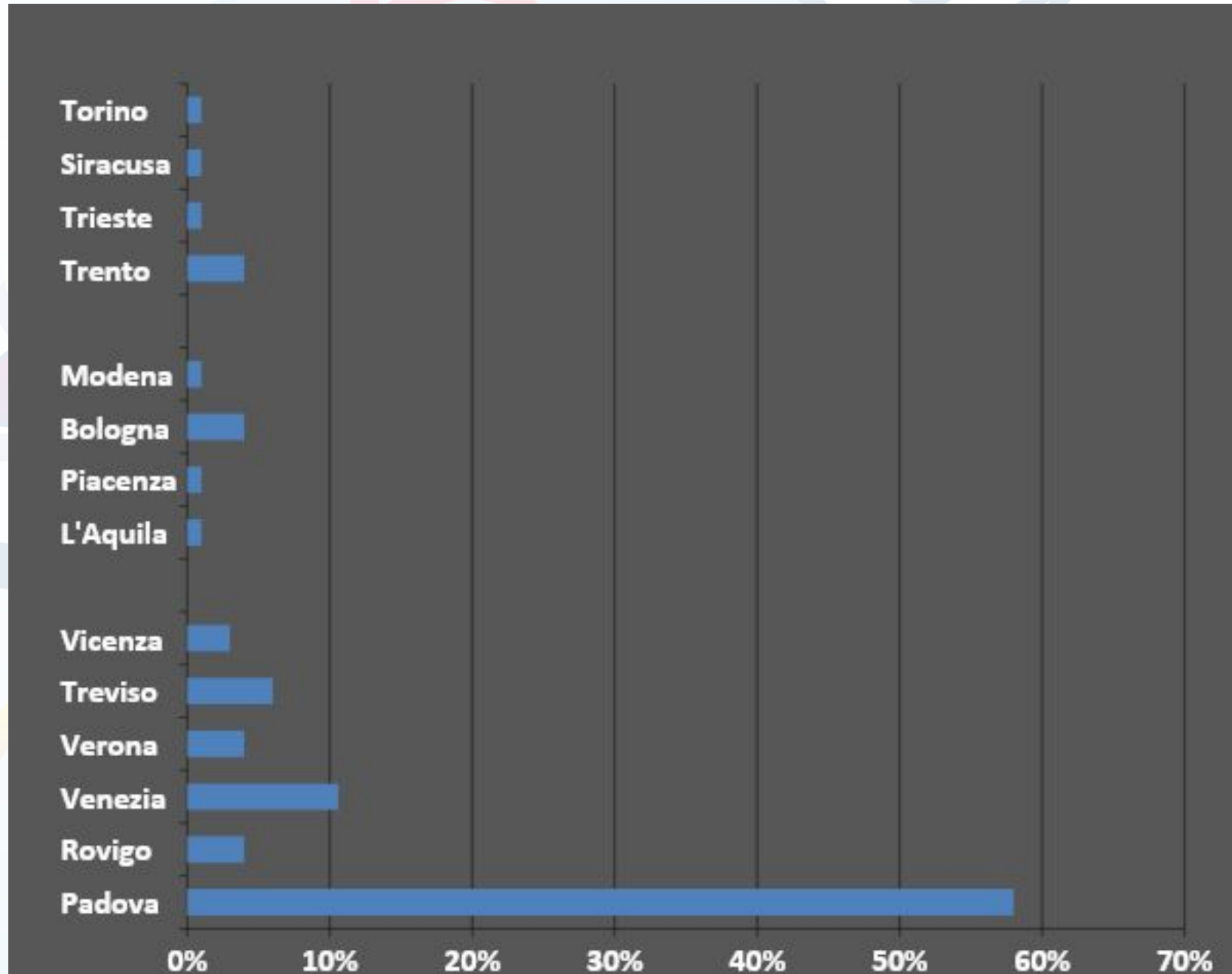


User graphs 2022

CSMR

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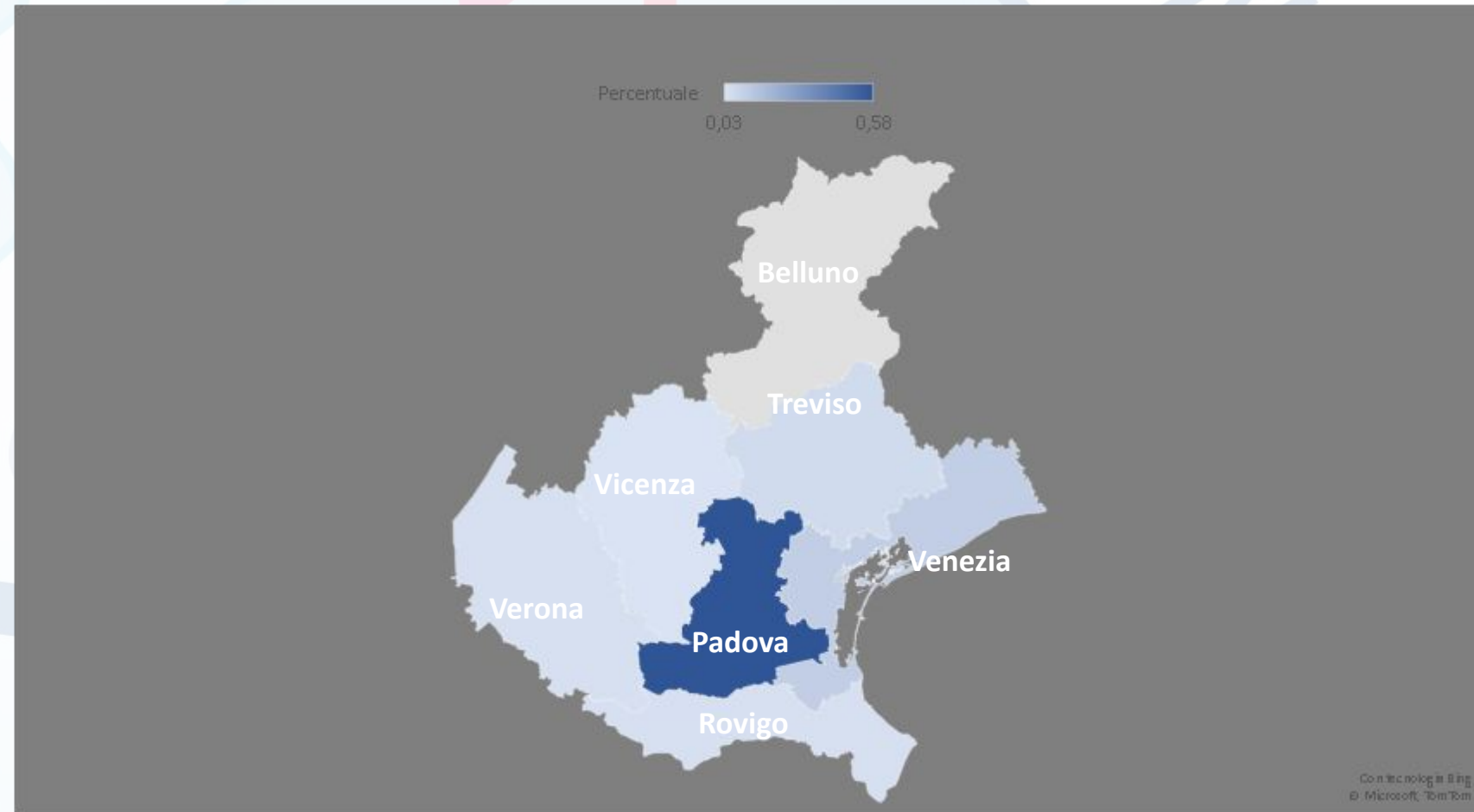
Cities with more contacts



Situation in the Veneto Region

User graphs

Users received 2023



Email contacts	Contact by Phone	Sporadic contacts	Total contacts received
424	253	450	1127

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Organigram - persons involved

<u>Activities</u>	<u>Person</u>	<u>Description</u>	<u>Location</u>	<u>Stakeholder</u>
First telephone reception	Cristina Bolzonella	Telephone answers 24/24	mobile	Aulss 6
First physical reception	Dott.ssa Roberta Bresciani	Mondays and Thursdays 9 a.m. to 2 p.m. or by appointment	Aulss 6 Via dei Colli	Aulss 6 for-profit companies
Empowering, listening and life coaching the patient	Dott.ssa Giulia Volpato Dott.ssa Nadia Ferrarese	By appointment	«	
Psychological counselling	Dott. Michele Tricarico	By appointment	«	
Legal advice	Avv. Mugnone Marcello	By appointment		
Self-help group - monthly	Dott. Michele Tricarico	1 Saturday per month 10 a.m. - 12 p.m. (at COVID-19 online time)	«	
Family training	By the Association p63 E.E.C. Syndrome International Onlus and Staff	Organizzazione training incontro famiglie e pazienti	«	
Secretariat and Communication	KmZero.Eu A.P.S.	Organisation of patient and family events, press office.	Detached headquarters	

Business plan

Human resources	Hours	Net cost €	Self-financing	Co-financing
Secretariat	h. 20 settimana	400,00 month - 4.000 year		
Reception	h. 20 settimana	750,00 month - 7.500 year		
Empowerment	h. 6 settimana	600 month 7.200 years		
Psychological counselling	Su richiesta	price list 50 euro per hour		
Legal counselling	Su richiesta			
Self-Mutual Help Group No. 3 professionals		1.000,00 month - 10.000 years		
Press office		500,00 month - 5.000 year		
Management expenses		1.500 month - 15.000 year		
Total		48.700 €/year		



About us...

Announcement of the opening of the first Rare Disease Service Centre, a SOCIAL support point for patients and families of patients with rare genetic disorders.

https://m.facebook.com/story.php?story_fbid=2152626491638240&id=1551716115062617

<http://mattinopadova.gelocal.it/padova/cronaca/2018/05/12/news/malattie-rare-inaugurato-un-nuovo-centro-1.16827052>

<http://www.padovaoggi.it/salute/centro-servizi-malattie-rare-ospedale-colli-padova-12-maggio-2018.html>

<http://mattinopadova.gelocal.it/padova/cronaca/2018/05/10/news/malattie-rare-apre-a-padova-il-primo-centro-servizi-in-italia-1.16817685>

<https://video.gelocal.it/mattinopadova/locale/padova-inaugurato-il-primo-centro-servizi-sulle-malattie-rare/94588/95053>

http://www.youreporter.it/foto_inaugurazione_SPORTELLO_CSMR_Centro_Servizi_Malattie_RAre



Thanks to all for your attention!

Contacts:

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C.S.M.R.

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