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IPWSO

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Caring and living with Prader-Willi syndrome in Italy: integrating children, adults and parents' experiences through a multicentre narrative medicine research

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Abstract

Objectives Prader-Willi syndrome (PWS) significantly impacts health-related quality of life; however, its relational and existential aspects remain unknown in Italian clinical and social debate. The project aimed to investigate the impact of PWS on illness experience through narrative medicine (NM) to understand the daily life, needs and resources of patients with PWS and their caregivers, and to furnish insights for clinical practice.

Design and setting The project involved 10 medical centres of the Italian Network for Rare Diseases and PWS family associations and targeted underage and adult patients with PWS and their caregivers. Written interviews, composed by a sociodemographic

NARRATIVE MEDICINE DEFINITIONS

“Narrative provides the meaning, the context, the perspective of the patient.
It defines how, why, and in what way the person is ill”.

Greenhalgh and Hurwitz. Why study narrative medicine. *BMJ* 1999

“Narrative Medicine strengthens clinical practice with the narrative skills to recognise, absorb, metabolise, interpret, and be moved by disease narratives: [...] it helps doctors, nurses, social workers and therapists to improve the effectiveness of care by developing the capacity for attention, reflection, representation and affiliation with patients and colleagues”.

Rita Charon. Narrative Medicine: A Model for Empathy, Reflection, Profession, and Trust. *JAMA* 2001

AT THE ROOTS OF
NARRATIVE
MEDICINE:
INTEGRATING THE
THREE
DIMENSIONS
INDIVIDUATED BY
A. KLEINMAN

Disease

The **biomedical definition** of the condition.

Illness

The **subjective experience** of the condition, including feeling, thoughts, wishes, needs.

Sickness

The **social and cultural meaning** of the condition; it may influence how the patient reacts.

THE PRADER-WILLI SYNDROME

Prader-Willi syndrome is a rare genetic disorder characterized by hypothalamic-pituitary abnormalities associated with hypotonia in the neonatal period and the first two years of life and the onset of hyperphagia, which can lead to the risk of morbid obesity in childhood and adulthood, learning difficulties, behavioral disorders or psychiatric problems. The syndrome occurs in both males and females and is the most frequent rare genetic disease causing obesity, with a prevalence of 1:10,000 to 1:30,000 cases.

PRADER-WILLI SYNDROME



Care must be **comprehensive and multidisciplinary**, with a team of endocrinologists, nurses, psychologists, educators, psychomotor therapists, social workers and general practitioners.



Early diagnosis, early multidisciplinary therapy and treatment with growth hormone (GH) have significantly improved the quality of life of these children.

PRAXIS PROJECT

Objective: to shed light on a condition that is not sufficiently well known in our society, confined to the scientific world of rare diseases by collecting the **narratives of people with Prader-Willi, family members and health professionals.**



Understanding and responding to the real needs of families (**caring**)



Implementing organisational recommendations to provide more efficient services for families (**curing**)



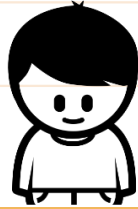
Identifying and minimising the stigma associated with living with and caring for people with Prader-Willi

Praxis: since October 2018 to July 2019

Praxis during the COVID-19 pandemic: Sep 2020 - Nov 2020

The study was unconditionally sponsored by Sandoz

RESULTS



107 compositions by
people with Prader-
Willi syndrome
(children, youths ,
adults)

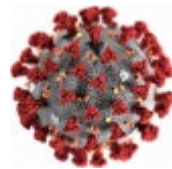


112 family
members
narratives



22 parallel charts
by health care
providers

241 Narratives (155 expected),
78% extra participation



15 compositions
compositions by
people with
Prader-Willi
syndrome

26 family
members
narratives

18 family members
narratives

59 Narratives during the Covid-19 pandemic

THE POINT OF VIEW OF THE PERSON WITH PRADER-WILLI

Children and youths with Prader-Willi

Gender	Boys 74%	Girls 36 %
Average age	14 years	13 years
	Mean 14 years- Min 7; Max 18 anni	
Study title	Primary school: 43% Secondary: 43% High School 14%	
Age at diagnosis	2.33 years	Min: at birth Max: 8 years
Age when informed of the diagnosis	8.36 years	Min: since ever Max: 15 years
Geography	North 13%; Centre 19%; South 69%	

The variation between North, Centre and South among children and adults is a consequence of the types of centres chosen.

Adults with con Prader-Willi

Gender	Men 44%	Women 56%
Average age	26 years	31 years
	Mean 29 years (Min: 19 anni Max: 48 anni)	
Study title	Primary School 6% Secondary: 30% High School 64%	
Civil status	Single 99%	
Work/Study	Work 56%; Study 11%; Does not work 33%	
Age at diagnosis	4.51 years	
Age when informed of the diagnosis	13.37 years	
Geography	North 58%; Centre 18%; South 24%	

Health and social care lead to a high school diploma in 64% of people with Prader-Willi.



WHAT I LIKE TO DO...

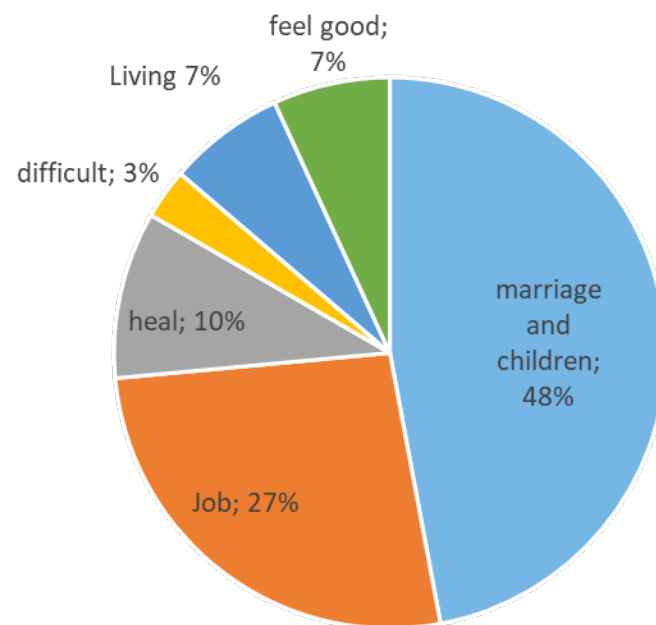
Arts: 20%, Home activities: 18%, Sports: 20%, Social hobbies: 12%, Pets: 8%, Food: 4%, Religious engagement: 4%

People with Prader-Willi have a wide range of interests: sporting activities, some of them even at a competitive level, *'swimming, I'm very proud, I've won medals'*, zumba dancing, a *passion for nature* and for animals as a form of pet therapy. Enjoying being out in the countryside and gardening. At home, the activities involve sport, the use of consoles, but also manual work.

- Extraordinary visual-spatial intelligence.
- Musical intelligence.
- Natural intelligence
- Spiritual intelligence



FUTURE PROJECTS



Marriage and children	«I am getting married with my future husband and my biggest dream is to become a mother one day.»; "I would like to get married, have a child and be happy"
Job	«working in the "special kids"project" in pizzeria, taking orders, job is important for me. With the help of the psychologist I keep the little devil away."
Heal	«heal», «I hope a drug will be found to limit nervous hunger and increase the sense of satiety."
Feel good	«I hope I feel good», «being well with my parents, sister and grandchildren".
Living	«living day by day, letting destiny and loved ones change my life "
Difficult	"I think it will be harder and harder", ""there will be no future""

MULTIPLE INTELLIGENCES

Linguistic and reflective intelligence on words;

Logical and mathematical intelligence;

Musical intelligence;

Visual-spatial intelligence;

Kinesthetic intelligence, body intelligence;

Interpersonal, relational intelligence;

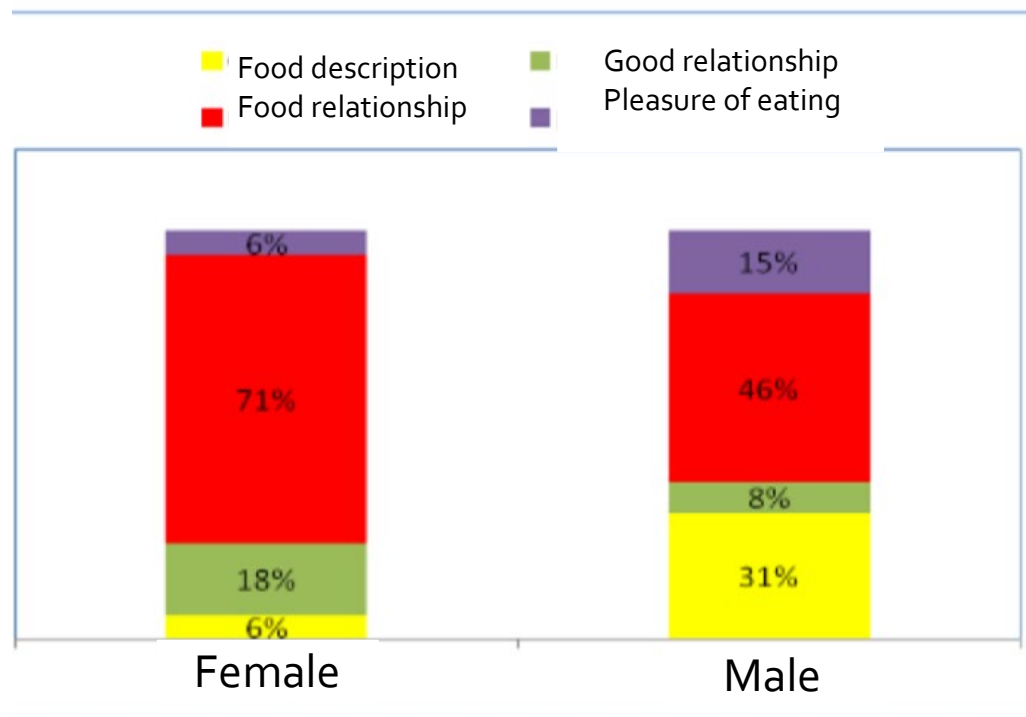
Introspective intelligence, about emotions and moods;

Natural intelligence, the relationship with nature;

Existential intelligence, reflections on good and evil;

Frames of minds, Howard Gardner, 1984

THE FOOD RELATIONSHIP

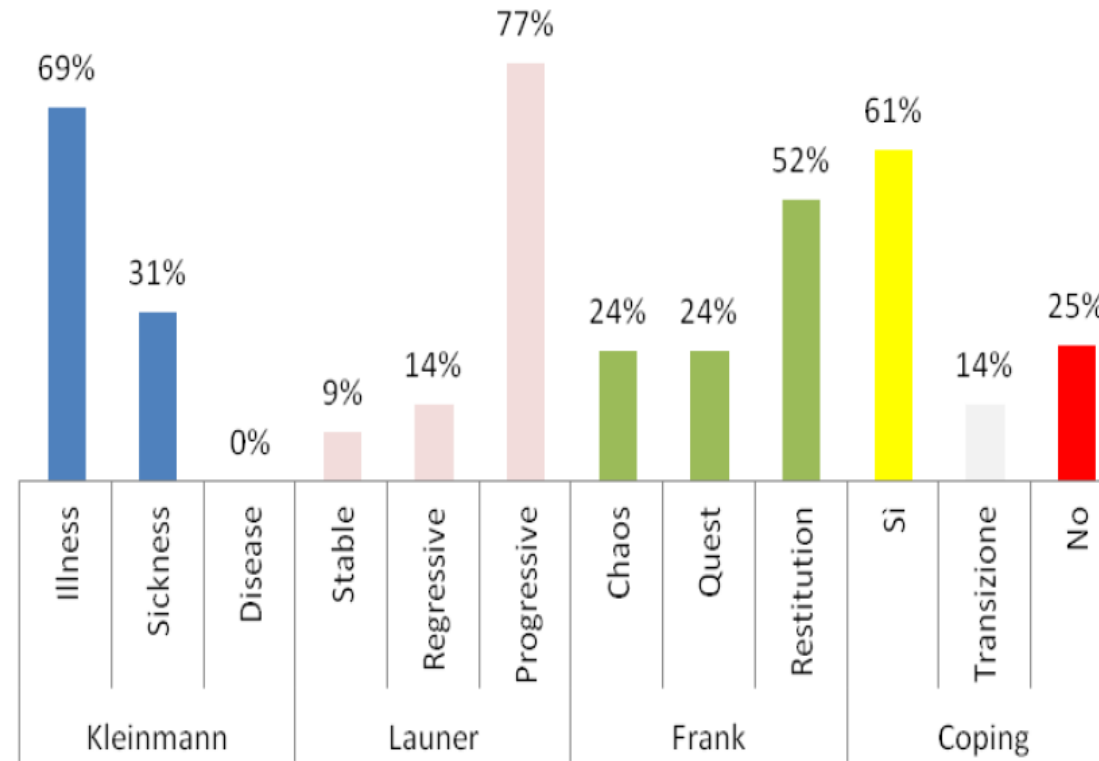


Children and adults with Prader-Willi tend to be in a position of "Restitution", i.e. they follow the diet required of them with discipline (and effort), with the expectation that their health will be restored to them in return ("I know eating hurts so much with my illness, which is called Prader-Willy syndrome").

Women report a more complex relationship with food and talk about HOW they eat, while men focus more on WHAT they eat.

Young people write about their strategies to avoiding to eat, or to thinking of the food: go out, and walk, stay in contact with nature, practice a lot of sports, putting themselves in a «discovery» mode- «quest»

CLASSIFICATIONS OF NARRATIVE MEDICINE



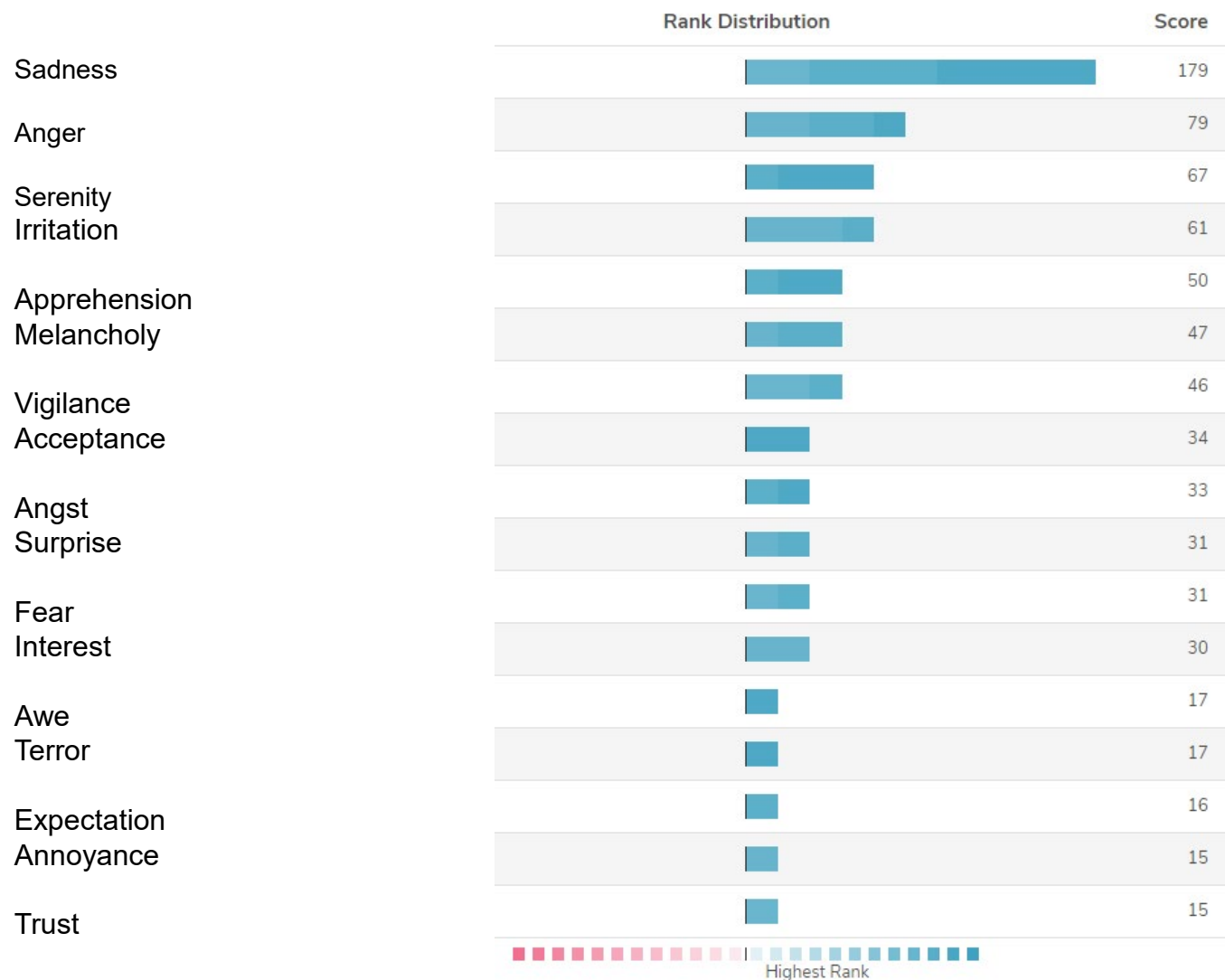
Prevalence of "progressive" narratives - 77%, i.e. stories that have a progression towards desires and wants (even present in 97% of cases). 14% are of people in regression, where there is a retreat in terms of energy, willingness and confidence ("when I am old I will stop caring and die").

The 'illness narrative' (69%) prevails, i.e. the free stories, where the person expresses himself without filters, generously opening up to the story, touching on different aspects of his own emotions, thoughts, desires, episodes. Disease narratives are missing (0%), i.e. those in which the focus is on the disease and its clinical aspects. Sickness is present, the difficulty that go through in 31% of the narratives of living with Prader-Willi. Stigma: "...I've never had friends in my life because everyone thinks I'm sick and contagious..."; "Sometimes I'm sad because I can't have friends"; "They don't call anyone, they're mean, they don't love me because I have the syndrome"; "); there are episodes of discrimination at school and teasing by classmates.

BEFORE AND DURING COVID- 19: 16 PEOPLE'S NARRATIVES

My life before Covid-19	My life during Covid 19 age
<i>"full of commitments linked to school activities in the mornings, rehabilitation therapies, sports and socialising activities, weekends linked to exercise and outdoor life, always socialising".</i>	<i>"school stopped, sporting and rehabilitation activities were suspended, online I was unable to learn and get active, tension at home was high, but no one took away my desire for an extended walk with the dogs in the open air. I've gained a few kilos and the desire to escape to a countryside far from incivility is strong in me."</i>
<i>"I played, I walked, I went to school".</i>	<i>"no more school but playing and walking"</i>
<i>"Before covid19 I was doing many things and my life was full."</i>	<i>"The virus stopped everything. I stayed at home for many days without doing anything and I gained weight."</i>
<i>"I had work experience in the school secretariat, went to the swimming pool without having to book my attendance, went out freely and saw the rest of my family freely. "</i>	<i>"I had to take measures for my own health security and for the rest of my family and friends."</i>
<i>"I was very cheerful before covid came along I went out with my friends I went scouting I went to school and I went out a lot with my parents".</i>	<i>«then covid 19 arrived and we were locked at home so as not to make a crowd, the schools were closed and we studied with distance learning with the pc, tablet, I did the tests online"</i>

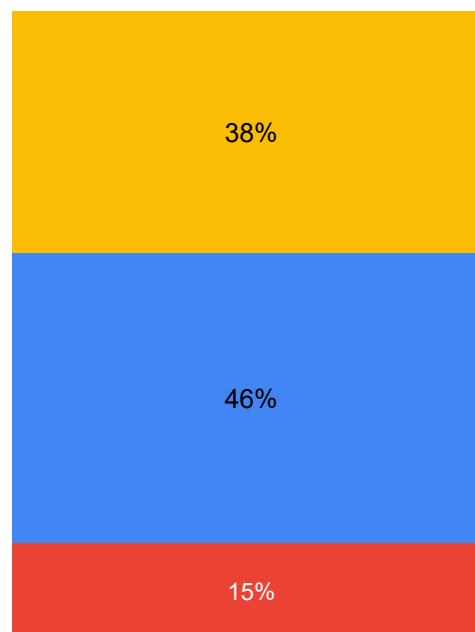
EMOTIONS DURING LOCKDOWN



PHYSICAL EXERCISE AND BODY DURING LOCKDOWN

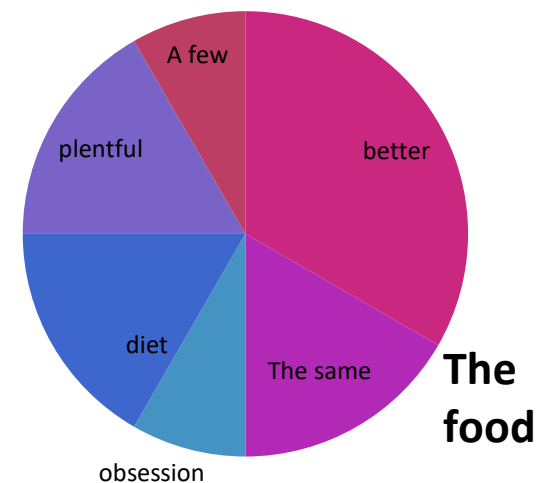
PHYSICAL EXERCISE

- I did not practice
- I practiced less than usual
- I practiced the same



THE BODY

"the covid arrives and... since I worked every other week with my husband every day they spent time doing gymnastics they did the landing 21 times and then the garage space about 70 meters back and forth for another 20 times: we took it as a game a competition between mother and daughter."



CAREGIVERS— FAMILY MEMBERS

Kind of relatives	Mothers 70%, Fathers 27%, Brother/Sister 3%	
Gender	Men 27%	Women 72%
Age	Mean 48 years - Min 20; Max 61 anni	
Study title	Primary school 4% Secondary school 14% High School 55% University graduation 14%	
Civil status	Married 86%; Single 2%; Divorced 10%; Widow 2%	
Geography	North 21%; Centre 29%; South 50%	
No. Inhabitants	<25.000 (42%); 25.000-50.000 (15%); 50.000-250.000 (12%); Beyond 250.000 (31%)	
Working situation	Employee 53%, Free lance 15%; Not working 28%; Retired 2% Student 2%	
Age at diagnosis	1.54 yearsr Min: 14 years Max: at birth	

138 family members left their narratives, more than double the 60 expected narratives, indicating a willingness to participate and be heard.

Women, generally mothers, told their stories in 72% of cases, men, generally fathers, in 27%. Although the latter figure may seem low, compared to other surveys conducted through narration, the response of fathers in recounting their experience is significant.

COMMUNICATING THE DIAGNOSIS OF PRADER-WILLI, CRUELTY OF DIAGNOSTIC LANGUAGE?



- *"“.. told us that she was suffering from Prader-willi, a very rare disease whose characteristics were a compulsive search for food that would lead to obesity, a very severe mental retardation, which would not exceed 6 years of age...”"*
- *The phone rings: "Is this Mr? This is confirming the diagnosis, your daughter has Prader-willi syndrome." "Excuse me, but what is this disease?"*
- *"I'm sorry but I have to inform you that your daughter has a rare generic disease called Prader-willi, an alteration of chromosome 15. Unfortunately we know almost nothing about it, the only thing we know for sure is that the patients have no hunger control and the big problem is obesity with all its consequences."*

There are many Prader-willi with different severities: how to communicate without resorting to stigma?

THE BURDEN OF ILLNESS



*"we didn't even go out for a pizza anymore",
"guarded on sight", "constant struggle to
make the grandparents understand the
situation".*

*"it breaks my heart to see my daughter when
she meets someone with ice cream or a cake".*

*"I avoided taking her to places where she
could see food, I took the dogs to eat in her
absence, I made her eat alone and never with
company".*

"our way of life has changed"

"I need to unplug from time to time."

*"For me it's become an obsession, I'm like a
jackhammer, you can't eat that, that's
weighed right?"*

*"You need a lot of patience, maybe more than
Job's."*



THE WORK RELATIONSHIP



With respect to work, from the word cloud, adding together the words clerk, employee, clerical, administrative, parents mainly carry out office work and it is a professional situation that, if chosen, can be reconciled well with the need to be near their son/daughter with Prader-Willi.

62% of parents had to change their profession after the birth of their son/daughter with Prader-Willi. More than a third gave up their job. In terms of gender, while 67% of fathers manage to keep their professional situation unchanged, it is women who make the most sacrifices, with only 37% managing to keep their professional status.



THEN COVID CAME: 22 CAREGIVERS SHARED THEIR STORIES

*"We found ourselves isolated. My husband works outside the region and only comes back at weekends. **The total lack of possibility to relate with others, if not via zoom, brought my son to pathologically bind himself to me considering me the only reference point.** The psychiatric situation has precipitated without the possibility to receive any help except for the psychopharmacological one, which was not enough.*

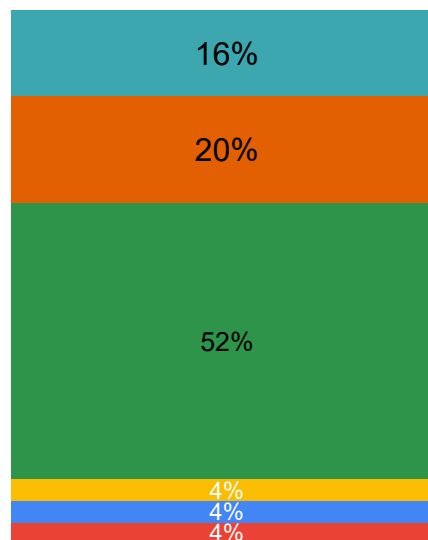
*"sports and social outings were missing, but they were replaced with video coaching, video family, video calls. **Despite being at home and in isolation, we tried to maintain our habits as much as possible.** We succeeded also thanks to the formidable support of the Sports Association and the Sfa Centre that M. attends."*

"we shared most of the time trying to spend it actively".

*"Covid19 arrived and all social activities were interrupted... school... sports and friends. we tried to maintain social contacts using available technological tools (video calls, phone calls, chats) and as soon as the lockdown conditions subsided, with social distancing and outdoors contact with friends resumed. Now S. has started secondary school and the spread of the virus is accelerating. **Some new worries appear about how she will be able to cope with a school year with distance learning when not all the teachers are yet known and the class group has hardly had a chance to form.***

ACCESS TO HEALTH CARE FACILITIES DURING PANDEMIC IN 2020

- Yes
- No planned visits
- Visits were postponed
- Visits were cancelled
- I cancelled visits or planned tests



"as they were only check-ups, no harm was done".

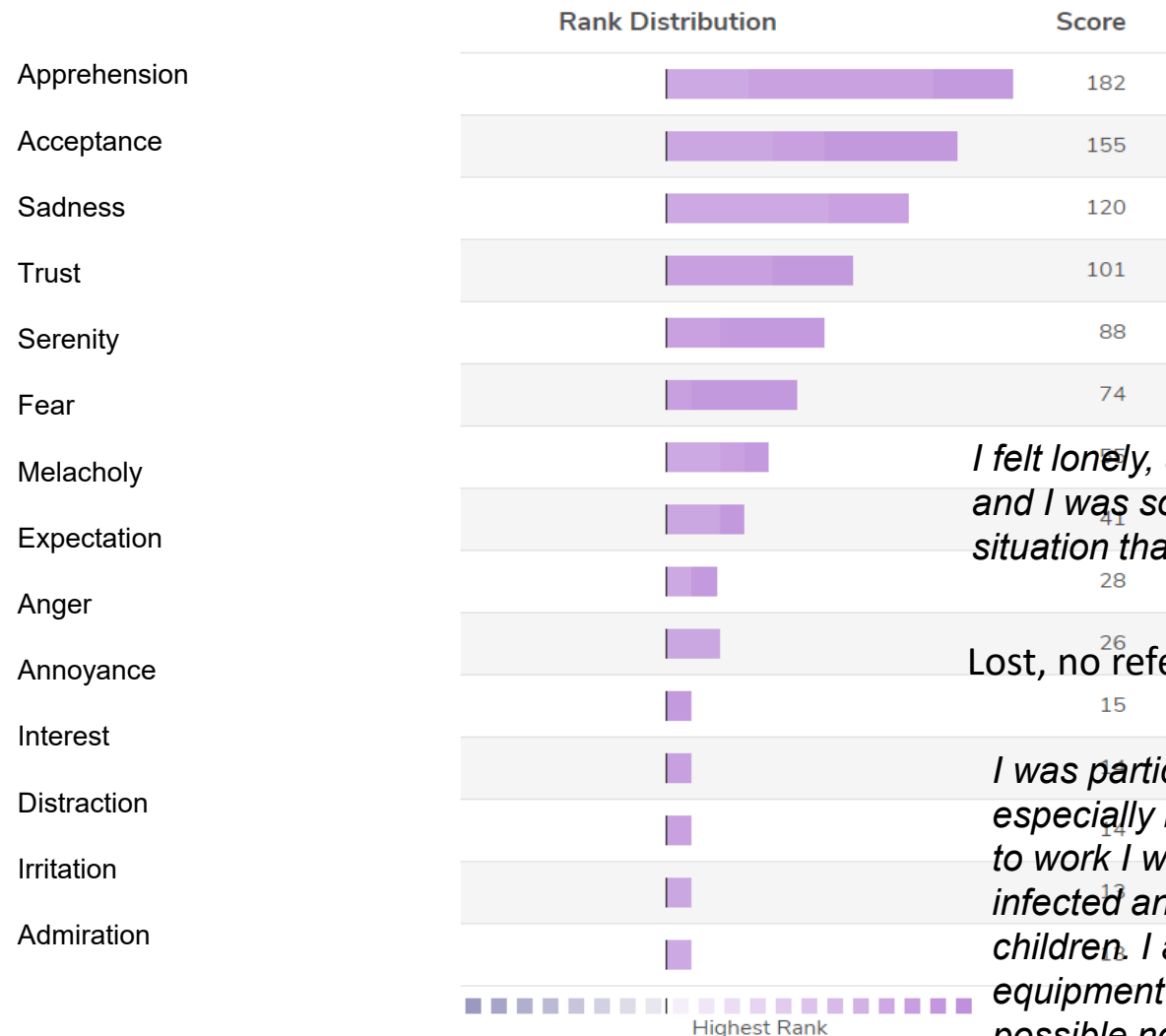
"medical disinterest"

"There were no major difficulties in managing the boy with Prader-Willi during the lockdown period".

"My son was a bit bored and nervous".

*"All in all Positive period, he accepted all the various restrictions, Masks , Disinfectant Gel and Spacing. Fortunately our living area is in the open countryside , the green UMBRIA , so with ample opportunity for mobility in the surrounding nature . There have been a few moments of discomfort, in which the reaction of our son was more mature, urging us sometimes shouting at us to keep their distance and wear masks. Even **during several walks, at the sight of a car, his reaction was to close his mouth so as not to ingest the virus, forcing those accompanying him to do the same thing**".*

I FELT (THE CAREGIVER) ...



I felt lonely, abandoned, sad, thoughtful and I was so afraid of the whole situation that had arisen overnight.

Lost, no reference points

I was particularly anxious and worried, especially because by continuing to go to work I was afraid of becoming infected and passing the virus on to the children. I also used protective equipment at home, trying as much as possible not to burden the children with the situation. At that time the news was bombarding me and I was very confused.

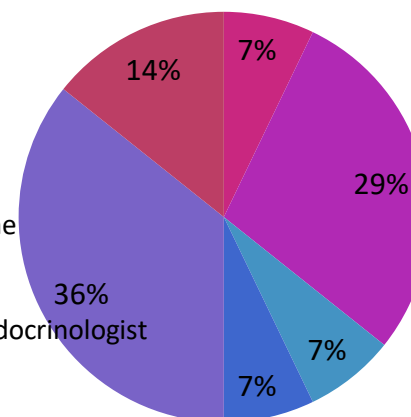
THE POINT OF VIEW OF HEALTH CARE PROVIDERS

Health care provider of Prader-Willi people		
Gender	Men 32%	Women 68%
Age	Mean 46 years - Min 24; Max 62 years	
Years of experience caring Prader-Willi	Mean 14 years – Min 1; Max 28 years	

Principal facts:

- early treatment importance
- multiprofessional and multidisciplinary approach

Dietician
Endocrinologist
Physical Medicine
Nurse
Pediatrician
Pediatrician/Endocrinologist

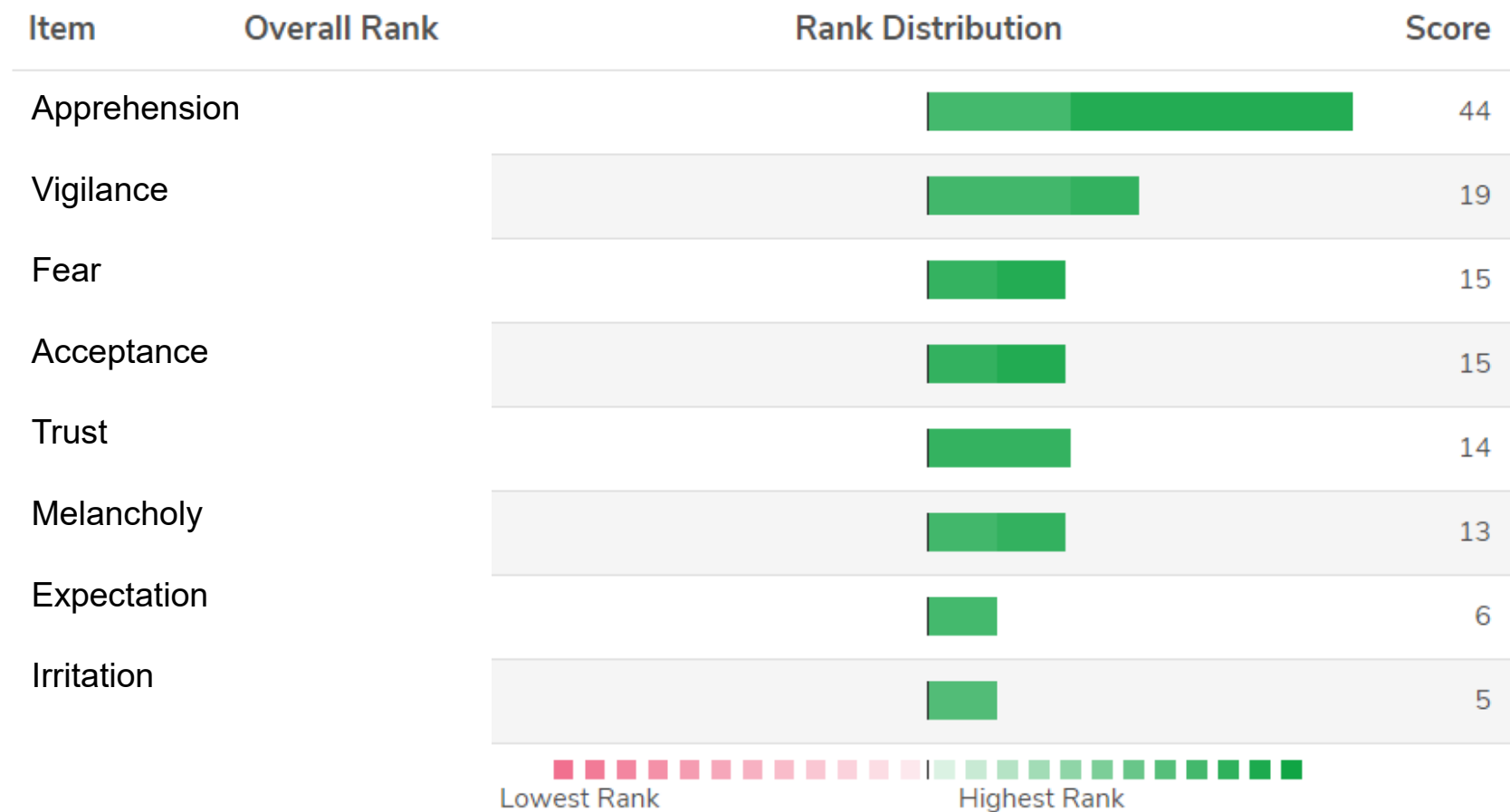


■ DIETISTA
 ■ Endocrinologia
 ■ medicina fisica e riabilitazione
 ■ O. S. S.
 ■ Pediatria
 ■ Pediatria/Endocrinologia

Health care providers during iCOVID		
Hospital	Hospital 36%, Academic Hospital 36%, IRCCS 28%	
Gender	Men 21%	Women 79%
Age	Mean 56 years - Min 29; Max 70 years	

22 health professionals reported their experiences, 13 during Covid-19.

I FELT (THE
HEALTH CARE
PROFESSIONAL) ...



THEN COVID CAME...

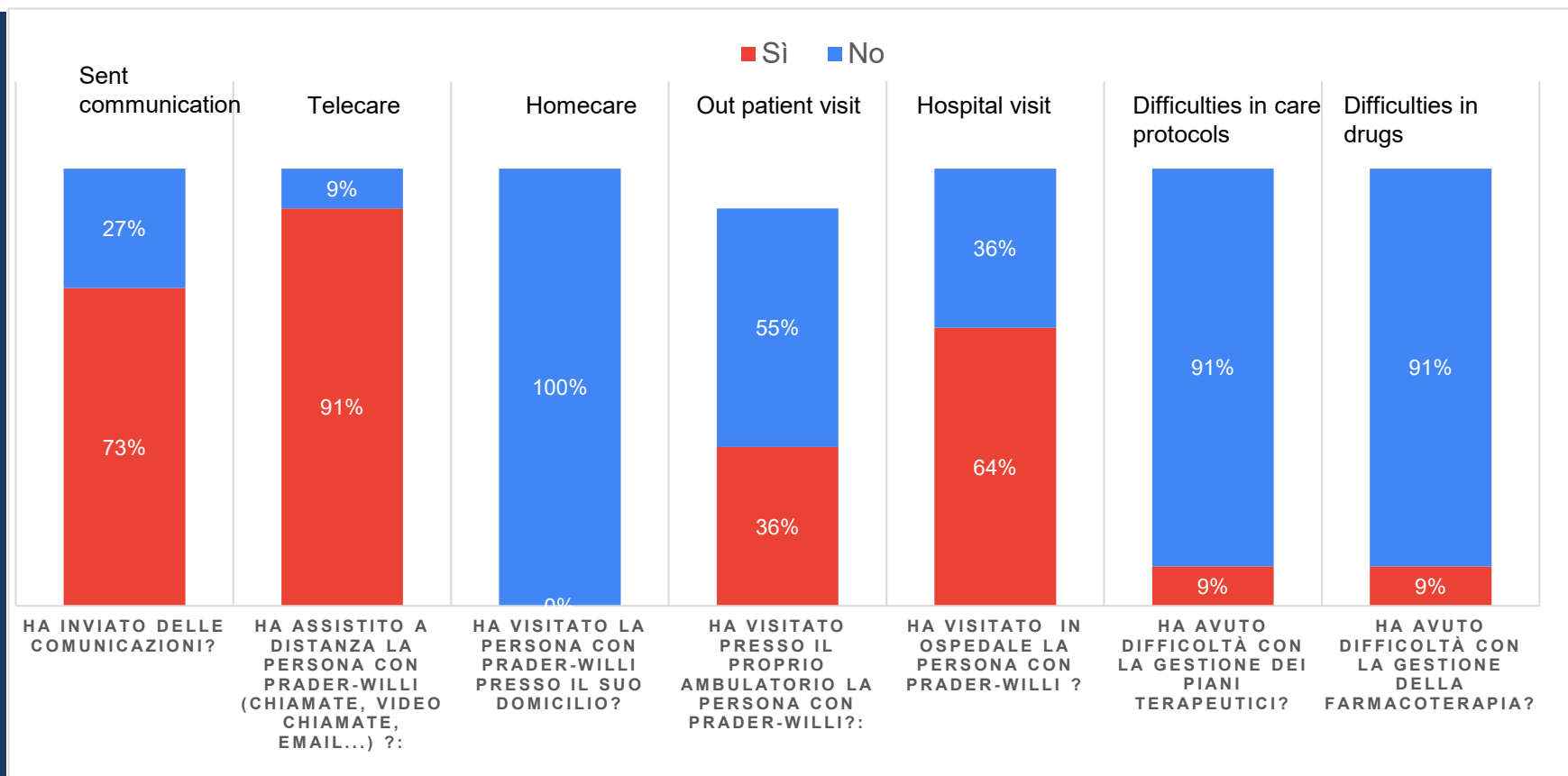
*...everything was blown up, **hospital shifts were revolutionised with increased workload and all scheduled activities were blocked.** The result was a further increase in online and telephone activity, in addition to that already provided routinely..*

*During the lockdown period **I only went out when justified and for special reasons.** Hospital activity was eliminated for obvious reasons.*

The boys stayed indoors and had serious problems with food and weight management.

*"We became an outbreak so everything turned upside down, we had some subjects with PW positive to covid, but fortunately they overcame the moment, I can say that they were really calm because they were looked after by all of us, the ones who were not calm were us operators, they were the ones who supported us. **We started with the video calls, the parents send their consent to the video, and I carry out the control visits and send the GH treatment plan**".*

EMERGENCY MANAGEMENT



"We closed the clinics and then reopened them to a few patients, only the most urgent cases.... Pre-triage, devices for us and for patients everywhere.... In the ward admissions only for urgent cases and after negative swab after standing in grey air."

"I have maintained care support for all PWS patients I follow, with drastic reduction of admissions and outpatient visits (to zero) and hypertrophy of telematic and telephone support."

"I have heard from many patients on the phone and we have exchanged a lot of information x email."

FINAL CONSIDERATIONS

- This study allowed **18% of the entire population** of people with Prader-Willi in Italy to tell their stories.
- People with Prader-Willi experienced **with great enthusiasm the request to tell their stories and to express themselves** by telling moving stories, looking for formulas to build well-being
- Families of people with Prader-Willi and health professionals welcomed telling their stories as a great opportunity to express **their own experience and functional and dysfunctional aspects in the care and social services in Italy**
- The relationship with **food in the pre-COVID era was easier to manage** as a result of the many distracting activities of children and young people (school work, day centre, sports, walks, friends, theatre, etc.)
- **62% of family members had to leave or change jobs, most of them women**, in order to devote themselves to the person with PW, the relationship with the Associations and the PW federation as a point of reference is fundamental
- Treatment with **growth hormone and other drugs are considered useful** and necessary by people with PW and family members with PW caregivers
- The way in which the first diagnosis of PW is communicated needs to be taken care of - **memory goes to the cruelty of "branding" and "predictive" words of future incapacity**

FINAL CONSIDERATIONS: THE IMPACT OF COVID-19 IN THE CARE OF PEOPLE WITH PW

- COVID **suspended time and outside activities**, which are very powerful therapeutic factors in helping the relationship with food. All blocked, with great boredom reported by people with PW.
- The relationship with food, immediately available in the fridge, becomes more difficult. **Caregivers are called upon to invent creative solutions** to create distractions.
- There is a mismatch between what is reported by the relatives of people with PW and the health professors on the access to care: **the caregivers have postponed the care, while the professionals have been present in the places** where they were received and have tried to make virtual through telemedicine their proximity to people with PW, in order not to interrupt the therapeutic relationship.
- In the emotional side, **sadness prevails in people with PW while in family members and professionals, vigilance prevails.**
- The COVID is metaphorically dismissed as **a monster, i.e. an aberration of nature**, rather than as an enemy, which recalls a situation of great conflict.

FINAL CONSIDERATIONS

- The testimonies of people with PW are proof of the limits of words such as "mental retardation" "reduced cognitive abilities" and instead open fronts **on other intelligences from visual-spatial to musical, natural and existential with creative solutions.**
- The **virtual can be the system for maintaining care relationships**, and a source of family closeness where smart working can be an opportunity to prevent interruptions and major changes in the workplace.
- What worked before the pandemic should be transferred to the homes and freedoms that people with PW and their family members must have: **opportunities to exercise in appropriate centres, to go for walks, more engaging teaching through gamification and less didactic;**
- **Health professionals in collaboration with social services can create an ecosystem of networks of study and collaboration**, to advance scientific and social research, achieve new forms of communication from the moment of diagnosis to these forced times of staying at home.