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Caring and living with Prader-Willi syndrome: integrating children's, adults', and parents' experiences through Narrative Medicine

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Abstract

Objectives. Prader-Willi syndrome (PWS) significantly impacts Health-Related Quality of Life (HRQOL). However, its relational and existential aspects remain unknown in Italian clinical and social debate. The project aimed to investigate the PWS impact on HRQOL through Narrative Medicine (NM) to understand experiences, needs and resources of PWS patients and their caregivers, and to furnish insights for professionals' daily practice.

Design and setting. The project involved ten medical centres of the Italian Network for Rare Diseases and PWS Family Associations and was addressed to PWS underaged and adult patients and their caregivers. Written interviews, composed by a sociodemographic survey and a narrative, were collected through the project's website. Three dedicated illness plots structured narratives to encourage reflection, employing evocative and open words to facilitate individual expression. Narratives were analysed through NVivo software. Researchers shared the results in a validation meeting with the project's Steering Committee.

Participants. Twenty-one children and adolescents and 34 adults with PWS joined the project, as well as 138 caregivers. A PWS diagnosis or the caregiving of a PWS patient older than five years represented the eligibility criteria, as well as the willingness to share illness experience by writing and the ability to communicate into Italian.

Results. The narrative analysis led to understand PWS social and relational issues, concerning: diagnosis and current management; PWS daily experience and social context; PWS implications in the working sphere and participants' future perspectives. Narratives showed that PWS management crucially impacts on relationships and work-life balance, also revealing that social stigma is still present.

Conclusion. The project represented the first effort to investigate PWS impact on HRQOL in Italy through NM, simultaneously considering the perspectives of PWS patients and their caregivers.

Findings indicated that a multi-professional approach is fundamental to ensure adequate treatment and provided elements for its improvement.

Article summary

Strengths and limitations of this study

- Inclusion of PWS patients' perspective within the project
- Narrative Medicine approach
- The participants did not equally represent Italian different geographical areas
- Among PWS patients, researchers included only those able to write

INTRODUCTION

Prader-Willi Syndrome (PWS) is a rare genetic condition caused by an absence of functioning paternal genes on chromosome 15 in the 15q11-q13 region:[1] about 65-70% of the cases are due to the deletion of this region, 20-30% are caused by a maternal uniparental disomy (UPD) of chromosome 15, and most of the remaining 2-5% have an imprinting centre defect or unbalanced translocations (~1%).[2] PWS occurs approximately in 1 in 10,000 to 30,000 births,[3] affecting both sexes and all geographic areas.[4]

Neonatal hypotonia, poor sucking and feeding difficulties characterise PWS in early infancy; dysmorphic signs (mild craniofacial abnormalities, small hands and foot, kyphoscoliosis), multiple endocrine abnormalities (GH/IGF-I axis dysfunction, hypogonadism, central hypothyroidism and central adrenal insufficiency) and developmental delay constitute other cardinal features of the syndrome.[5-7] Learning disabilities, maladaptive behaviours, and hyperphagia – leading to life-threatening obesity if uncontrolled – follow in childhood and adulthood.[3, 8] The mortality rate of PWS patients is higher than in the general population,[9] with a 3% annual death rate across all ages.

Behavioural issues are noticeable in PWS, including aggressive and obsessive-compulsive behaviours and skin picking,[10, 11] and patients present a higher risk to develop psychiatric illness reaching adulthood;[12] food-seeking behaviours are particularly complex,[13] significantly impacting patients' and caregivers' Health-Related Quality of Life (HRQOL). In particular, PWS caregivers – when compared to other families facing children's disability or complex condition – report a higher level of stress, more difficulties in coping with symptoms,[14] a higher caregiving burden,[15] and a lower HRQOL.[16]

The clinical picture of PWS patients substantially differs during the life span,[3, 6] while the prognosis is significantly conditioned by proactive interventions to prevent morbid weight excess.[9] There is currently no healing treatment for PWS. However, early diagnosis combined with multidisciplinary care favourably influences PWS course:[17] consequently, the diagnosis should be confirmed very early during the neonatal period,[18] also thanks to genetic testing development.[19] In this context, early growth hormone (GH) treatment has beneficial outcomes on height, body composition, endurance, and sense of well-being among others;[20-22] furthermore, early treatment with recombinant GH positively impact on PWS[23] patients' and caregivers' HRQOL.[24, 25]

PWS social, relational, emotional and existential aspects remain profoundly unknown with a poor debate within Italian clinical and social communities: World Health Organization (WHO) has stressed the importance of HRQOL research in leading clinical and social practice, also recommending using narrative research.[26]

The discipline of Narrative Medicine (NM), based on illness narratives,[27] pursues to integrate the disease-centred approach, concerning clinical aspects, with the illness-centred and sickness-centred approaches, respectively focusing on the individual experience and the social understanding of a specific condition[28] and both often neglected by the scientific community. NM areas of

application range from the clinical practice to the therapeutic path design, education, and research.[29] In research, narratives show possible interventions on a specific condition through the integration of all perspectives involved in the pathway of care.[30, 31] Combining Evidence-Based Medicine (EBM) and NM provides clinicians methods to strengthen clinical practices with narrative competences.[29]

NM research addresses the individual experience when coping with distress caused by clinical conditions: it allows to understand profound experiences, needs and values of all actors involved in the care pathway.[27, 32] Scientific societies, healthcare facilities, and patients associations have increasingly employed results deriving from NM research to improve the organisation and efficacy of healthcare services and generating sustainability.[26] Moreover, efficacy becomes quality of care for patients and their social and relational contexts.[27]

The NM project "PRAXIS: Prader-Willi Excellence in Care with Story Taking" aimed to investigate the PWS impact on HRQOL, employing narrative analysis (a) to understand the experience, real needs and personal resources of people with PWS and their family caregivers from diagnosis to current management, and (b) to furnish insights to help professionals to use NM findings in daily practice within a multidisciplinary and multi-professional approach.[17]

METHODS

Patient and public involvement

The project was conducted in Italy between October 2018 and July 2019, as a part of broader research addressed, on the one hand, to people with PWS and their caregivers, and on the other hand, to professionals working with PWS. Professionals underwent a webinar conducted by researchers from ISTUD Foundation to be trained in NM; parallel chart[27, 33] was identified as the most suitable NM tool to collect their narratives since it represents a room in which professionals can write their impressions in plain language as a supplement to technical and quantitative

reports.[27, 30, 33] Caregivers were informed about NM through three seminars – organised by the Prader-Willi Federation and the Prader-Willi Association of Lazio Region – in Lombardy, Lazio, and Sicily Regions. Participants with PWS were given the possibility to express by drawing if unable to write; however, some participants decided to maintain both the opportunities of expression.

Participants were recruited involving ten medical centres for paediatric and adult patients of the Italian Network for Rare Diseases (Supplement 1) distributed between different geographical areas (North, Centre and South Italy), the Italian Prader-Willi Federation, and the Prader-Willi Association of Lazio Region. A PWS diagnosis and the caregiving of a person older than five years with PWS represented the eligibility criteria, as well as the willingness to share illness experience by writing; thus, the ability to communicate into Italian was indispensable for the inclusion in the project. Participants were informed on the possibility to view the project's results once shared (into Italian) on the project's webpage www.medicinanarrativa.eu/praxis.

Data collection

Written stories of experiences were collected through the project's webpage; after that, raw and anonymous narratives were downloaded as a Microsoft Excel spreadsheet. A sociodemographic survey and an illness plot[27] constituted the written narrative, specifically designed for three different groups (Supplement 2): underaged and adult patients with PWS, and caregivers.

The three illness plots addressed common aspects: (a) diagnosis and current management of the condition, particularly focusing on strategies to food behaviours; (b) the daily living with PWS, concerning the relational sphere and social context; (c) the work experience and future perspectives. The plots guided narratives in a chronological order to identify evolutions over time. Evocative and open words were used to facilitate individual expression.[34]

The research tools were drawn up by a Steering Committee involving three endocrinologists, respectively from the Oasi Maria SS. Research Institute (Troina, Italy), the Bambino Gesù Paediatric

Hospital of Palidoro (Rome, Italy), and the Istituto Auxologico Italiano of Piancavallo (Oggebbio, Italy), and the researchers of ISTUD Foundation.

Ethical considerations

The project was performed according to the principles of the Declaration of Helsinki. Before their involvement, participants provided written informed consent after being briefed on the project's purpose and data confidential handling, according to the Italian Law on Privacy and the Safeguarding of Sensitive Data (D.Lgs n. 196/2003). Written informed consent to participate was obtained from parents or tutors for underaged participants, and from adult participants or their guardians, when appropriate.

The Ethical Committee of the Oasi Maria SS. Research Institute (Troina, Italy) approved the project in January 2019.

Analysis

We analysed the sociodemographic survey through descriptive statistics. No question was mandatory; we excluded non-responses from the statistical representation.

Anonymous narratives were entered into NVivo software[35] for coding and analysis. Researchers from ISTUD Foundation collectively coded ten narratives to assess consistency across team members. Afterwards, each narrative was coded separately and then reviewed during meetings to reduce bias in the interpretation of texts. Open interpretive coding was employed to identify and analyse any emerging topics; Kleinman's[28] theoretical framework was applied to narrative analysis to reveal illness- and sickness-related aspects in narratives, respectively concerning the personal and emotional experience of a condition and how is it perceived within the society.

Researchers shared the results in a validation meeting with the Steering Committee of the project.

All researchers use the SRQR Reporting Guidelines.[36]

RESULTS

Sociodemographic aspects

Twenty-one children and adolescents and 34 adults with PWS joined the project, as well as 138 caregivers. Table 1 summarises sociodemographic data of these three different groups.

Table 1 – Sociodemographic data of participants

	Minors with PWS (N = 21)	Adults with PWS (N = 34)	PWS caregivers (N = 138)
Gender			
Females	6 (29%)	19 (56%)	99 (72%)
Males	15 (71%)	15 (44%)	37 (27%)
Average age (yrs)	14 (7-18)	29 (19-48)	48 (20-61)
Geographic residence			
Northern Italy	3 (13%)	19 (58%)	29 (21%)
Central Italy	4 (19%)	6 (18%)	40 (29%)
Southern Italy	14 (69%)	9 (24%)	69 (50%)
Education			
Elementary school	21 (100%)	2 (6%)	5 (4%)
Middle school	//	10 (30%)	19 (14%)
High school	//	21 (64%)	76 (55%)
University degree	//	1 (1%)	19 (14%)
Marital status			
Single	21 (100%)	33 (97%)	3 (2%)
Married/cohabitant	//	1 (3%)	118 (86%)
Divorced/separated	//	//	14 (10%)
Widowed	//	//	3 (2%)
Employment status			
Student	21 (100%)	4 (11%)	3 (2%)
Working	//	19 (56%)	94 (68%)
Not working	//	11 (33%)	38 (28%)
Retired	//	//	3 (2%)

*Data presented as N (%) or mean (range)**

From diagnosis to the current management of PWS

Thirty-six per cent of caregivers reported that their children were diagnosed with PWS within the first month of life (Figure 1); however, 10% affirmed that the diagnosis occurred after the child's third year of life.

In narratives, 95% of caregivers focused on PWS illness- and sickness-related aspects (Table 2), while only the remaining 5% adopted a technical and clinical perspective for talking about the condition.[28] Disbelief, displacement, anger, and pain represented the most recurrent emotions expressed by caregivers, seeking to adapt to the situation and its criticalities. PWS patients – both

underaged and adult – described the condition only through its illness- and sickness-related facets, while technical and clinical references to PWS remained on the background.

Table 2 – Illness- and sickness-related aspects in narratives

	Caregivers	Minors and adults with PWS
Illness	*– It is not easy to live with a child with this syndrome, especially when she asks for food: I tell her, no, and she starts crying and getting upset, screaming and telling me things I do not understand, but I do not give up.* *– Broken dreams, the feeling that something had changed forever, that my life had changed. Me, so self-confident, so independent, suddenly feeling fragile, scared and alone, dreadfully alone and unable to react.* *– The first emotion was fear. I did not know how to manage his crises for food. I did not know what was happening to us, and every time we go to do the checks, we reopen the wound, with all its fears.* *– When my son was born, they immediately told me that something was wrong. It was terrible. I was young, and he was my long-awaited first child.*	*– I feel proud, disability does not scare me.* *– Sometimes I feel happy, and sometimes sad because I have this disease.* *– I feel very happy, sometimes a bit sad for my illness. But I am happy, because the Lord created me and I like me as I came into existence.* *– I feel a little sad for having this rare disease; there are many difficulties, especially not eating too much.*
Sickness	*– Concern intensifies, if I think that he will start school in September. I am worried about his level of tolerance, if he will become the object of insults and bullying.* *– Taking care of a boy with Prader-Willi syndrome means changing the lifestyles of the whole family, trying to combine the needs related to the disease with those of the family. It is a different journey, which could limit life experiences to those who do not have this kind of availability.* *– If I think of my child and her syndrome, I feel a great sorrow because I do not know if she will ever have a normal life.* *– We were often desperate, especially when we thought about our son's future. Then we learned to deal with problems as they appeared.*	*– Other people are bad. They do not care about me because I have the syndrome.* *– I do not feel up to the kids of my generation. I hate me, I want to die. Sometimes I would like to be a boy like the others, and always be happy.* *– Prader-Willi syndrome lasts a lifetime. It is difficult to communicate with others, even with parents in the family.* *– On the one hand, I feel different from others because I am disabled and hypotonic. On the other hand, I feel like them because I was lucky enough to walk, to see and to hear*

As shown in Table 3, focusing on therapeutic paths from caregivers' perspective meant addressing (a) relationships with professionals and (b) healthcare structures, both differing in specialisation and competence on PWS, and (c) needed or employed treatments, beyond diet.

Table 3 – PWS caregivers' perspective on therapeutic path

Professionals	*– Thanks to the specialist, we set up an appointment at the hospital, where we are currently being followed. The cases they deal with are many, and for us this is a*

	guarantee. At the human level they are unparalleled, available, smiling: all this allows you to spend more peaceful time, without feeling a burden. *– I started attending health workers, support teachers and incapable teachers, unprepared paediatricians, arrogant doctors, medical commissions, courts. I clashed with bureaucracy, absurd health protocols, illogical rules to get what my daughter was entitled to and her psycho-physical well-being.* *– I was disappointed by the professionals. They talked about my son and my life as something unmodifiable, fatal, hopeless. I decided to get as far away as possible. My son was not a syndrome, he was a child.*
Healthcare structures	*– We do not live in a big city, so we had great difficulty getting our daughter to follow suitably.* *– At the hospital they showed us the path we should have taken... on a medical level! On a therapeutic level, I had to resort to private professionals and centres.* *– Miles of road and hotel rooms distinguish the curative path that weighs on the family, both in economic terms and stress.*
Treatments	*– This part turned out to be better than expected. The only medicine is GH, which is simple and painless.* *– Psychomotricity, GH, speech and music therapy, psychological support and sport.* *– During adolescence, we had to resort to psychiatric drugs: this was a defeat for us, but it was necessary, our son had become unmanageable. There is little knowledge of the syndrome by psychiatrists.*

From caregivers' narratives, food-seeking behaviours[13] emerged as the most challenging within the domestic context. Caregivers were aware that feeding is the first treatment for people with PWS and sought strategies to face them. The 50% declared to have found a balance, while the other 50% still reported a problematic relationship with the food issue. Both underaged and adult participants with PWS showed to be aware of the importance of following a diet: whether positive or negative relationships with food emerged from narratives, where (a) the positive ones also represented the result of commitment and several strategies to manage food-seeking behaviours, and (b) food-seeking turned out to be related to emotions (e.g. anger). Table 4 shows the main elements emerged from caregivers' and PWS participants' narratives.

Table 4 – Relationships with food-seeking behaviours in narratives

	Caregivers	Minors and adults with PWS
Commitment	*– The first thing I did was going to visit a professor in Switzerland to find a diet as clean as possible that could give strength and good energy to my child. I decided to follow it too.* *– We started to have a different relationship with food, with more awareness of what we could and could not eat.* *– We tried to get the whole family accustomed to healthy eating as much as possible. I teach my son to read nutrition labels so he can choose the best one day. He already knows that he*	*– I have a good relationship with food, I can control myself, I eat legumes and soups, and pasta in the evening. I really like vegetables.* *– I follow a correct diet and I never steal food. If someone offers me something, I can refuse.* *– I follow a nutritionist's diet.*

	must not eat excessive sugar and fat; eat at precise times, but managing hunger is not easy.	
Strategies	*– We have almost always managed to keep it under control; we focused a lot on food education (salads, vegetables, no snacks). We gave her the possibility to choose from certain foods and she was therefore more satisfied, allowing us to eat differently.* *– Food is the main problem for my child but there are both positive and negative moments. So, we are tough if we need to be tough, but sometimes we make some exceptions.* *– Food is always in our mind, but we try to manage everything at the best. We try to live a life as normal as possible, and we try not to upset his habits too much. We give him some extras (he goes to parties, he goes out with some friends, he goes to eat pizza).*	*– I like to eat everything. I try to eat lots of vegetables, even if this doesn't fill me, and whole wheat pasta. When I play or do puzzle, I don't think about hunger. Mom tells me that if I want to eat more, I have to do more movement.* *– I try to organise my day according to fixed patterns, and I know I can eat at certain times.* *– In order not to think about food, I go for long walks, I do crossword puzzles and puzzles, I play tablet and computer.*
Criticalities	*– Unfortunately, food is an obsession and difficult to manage. Many times, I found my little child with hidden food.* *– The food issue becomes a daily challenge. Lunches and dinners are no longer quiet, and you live very anxiously. The kitchen is no longer a meeting place.* *– It is our conviction, a continuous struggle, day and night, the monster we have to defeat.*	*– I eat out of meal because I hate me. I do not want others, and I am hungry because my parents do not give me the right portions, and then I get fat and hospitalised, I steal food and go to the bar because I am for hungry. I did not eat as indicated by the diet.* *– It is a pain to see the others eating.* *– When I think of food, my eyes shine and when I see something to eat, I want it at all costs, and I cannot stop myself. I am always hungry, and I never get enough; when I eat too much, I feel sick. When they tell me that I cannot eat much, I get angry and nervous.*

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200 **Living with PWS in relationships and social context**

201 Thirty-six per cent of PWS caregivers described daily life at home as quiet; however, most of them
202 (64%) reported: fatigue (21%), chaos (6%), all-encompassing assistance (20%), and tested routines
203 to better manage food-seeking behaviours (17%). They try to maintain hobbies, interests and
204 outside activities, even with their sons/daughters with PWS (Table 5). Relationships external to the
205 family are hard to be preserved, imposing a radical change in social life. Indoor and outdoor
206 activities represented an essential tool for caregivers in managing both emotion patterns and food-
207 seeking behaviours: narratives showed that both underaged and adult participants with PWS were
208 aware of that. Sports, mind activities games, gardening and pet therapy were some of the most
209 helpful activities reported; furthermore, PWS patients appeared as dedicated to cleanliness and

routine activities. Both relational and activity spheres reported the influence of behavioural and emotional changes in daily life and familiar and social context; moreover, narratives addressed the strong presence of caregivers, as well as situations of social inclusion or exclusion.

Table 5 – Activities and relationships in narratives

	Caregivers	Minors and adults with PWS
Indoor activities	*– I like talking to my daughter, finding out what she likes to do and meeting her demands. I like to see her play with her friends and laugh, because she is very nice and charismatic.* *– I like being with my family; I'm a full-time mom. I would like to go to the gym and to be a woman like many others, but I do not have time.* *– In my spare time I am a musician.*	*– I like reading fairy tales and colouring.* *– Mom does not want me to go to the kitchen; I do puzzles or embroidery; I see Walt Disney cartoons. I read the newspapers. When I am sad or angry, I try to eat. Often, I quarrel with my mom. My dad says nothing because he makes me feel angrier.* *– I see the television, I hear the music, and I pray everyday. Every morning I make the beds and I lay the table while my parents are cooking.*
Outdoor activities	*– I reload being alone. I do yoga, I study Japanese, I sing in a choir. I do not leave my passions.* *– I like to spend my family life. I cultivate my hobbies thanks to my wife's patience. With her I share the passion for concerts and cinema. I also cultivate my personal passions: wine and the collection of stickers.* *– I would like to swim, to go for walks by the sea and the lake. I would like to have no responsibilities, schedules, limitations; I would like a little carefree.*	*– I play sports, especially football and swimming. I go for walks in company, alone or with my dog. I like going to see my favourite football team at the stadium. I play videogames and watch movies. I like going to see my favourite animal, the dolphin, at the dolphinarium.* *– I have so many hobbies: dancing, singing, painting. I like to do artistic activities, I like going to the cinema and to the theatre, I like reading crime novels, and I really like being with others and playing cards when the people around me do not fool me.* *– I like going out, shopping, swimming, listening to music, drawing, cutting and pasting, sticking figurines, making decoupage.*
Relationships	*– Some people have tried to understand the condition, others have disappeared. In this case, the syndrome helped me make selection among the people around me.* *– My father and my mother were as sorry as me and incredulous, unable to get over it. My sister and friends have been my support. My husband has shut himself up, present and absent at the same time, except for practical things.* *– The grandparents have been fantastic, available, always ready to help us. Friends did not understand our situation, and our roads soon split up: social life changes radically.*	*– My parents are the closest and most precious people. They are strict, but affectionate and loving. I am morbid with them and I always look for contact even if I make them angry. My brothers are also close to me. One is playful and the other protective, a little severe. With them I am serene and protective towards them.* *– At school I try to get close to the others, but they run away, they do not want me. Professors love me, even with some of them I feel I am different and not good. My parents: my biological mother did not love me, and my father hated me, my other parents love me so much.* *– The people next to me are my parents and my sister, and my personal support teachers. I do not like all my educators and my classmates because they spite me, but some of them are good.*

Work and future perspectives

Sixty-two per cent of family caregivers had to change their job after the birth of their child with PWS (Table 6): more than a third left their current work, 8% changed job to assist the child, another 8% requested for a part-time job, and 3% abandoned the perspective of a career. From a gender point of view, 63% of female (mothers) and 33% of male caregivers had to change their current job to adapt to the child's condition. Forty-six per cent did not talk about PWS in the workplace or did it just with their closest colleagues; 36% reported comprehensive behaviours, while 18% declared a lower understanding for PWS management necessities than for other diseases, such as cancer. From narratives, work also emerged as a positive personal resource. Fifty-six per cent of adults with PWS declared to have a job, mainly characterised by low complexity and repetitive operations and carried out in social cooperatives or centres, small companies with high corporate social responsibility, and family companies. Work is a source of pride and well-being and a distraction from food, but episodes of irritability and aggressive behaviours are also reported.

Table 6 – PWS and the working sphere in narratives

Adults with PWS	*– I am very happy because my colleagues are very good and they care about me, they are very close to me. I am happy because I like my job.* *– I deal with the press review, I answer by phone, I receive the people who come, and I do other jobs that the office managers assign me. If I am happy and motivated, I do not go looking for food. Only if I am nervous, I look for food. Now I have made so much progress and therefore I have done less.* *– I often have nervous attacks and I answer badly, offending with swear words those who make fun of me on how I do my job.* *– I work in a nursing home five days a week. I wash laundry, iron and fold the clothes of the guests. With me works a girl who follows me and teaches me many things. Every day I have a different person, it depends on the shifts. With them I feel good because they treat me like a normal person, and they congratulate me every time I work with them.*
PWS caregivers	*– I had the support and closeness of my colleagues, who gave me the priority for any personal needs, so to allow me to not leave my son and my wife alone. They were like a second family to me, they all contributed as much as they could.* *– I had to leave work. I worked at the factory eight hours and it was not possible to ask for a part-time job.* *– The job was a safety valve, a place to have a normal life.* *– At work, at first there were no changes, because I did not immediately say what had happened. I talked about it with a few trusted colleagues, who showed me great affection and solidarity.*

As for future perspectives, PWS family caregivers declared to hope to be present for a long time for their sons and daughters and to be particularly concerned about what will happen to them without

familiar support (Table 7). Caregivers also addressed social inclusion issues – such as social changes and openness – rather than clinical solutions to PWS. Adult participants with PWS showed a self-realisation perspective through work (27%) – as underaged participants do – and the desire to have a family (46%), as well as to recover from PWS (10%) and generally to live well (14%).

Table 7 – Future perspectives on living with PWS in narratives

Minors with PWS	*– When I grow up, I want to be a professor of endocrinology and study my syndrome.* *– I would like to be a vet because I love animals.* *– I would like to drive an ambulance and do a lot of jobs.*
Adults with PWS	*– I would like to start a family by adopting a child, because I have so much love to give, and being with my boyfriend. I hope to work in the same place. I do not know how to live without my parents, they help us a lot.* *– I would like to go living with the person I love the most in the world, having children and, above all, becoming a famous singer, and knowing all my favourite singers. I would like to get married and go to live in a nice house with a pool.* *– I hope that medicine will be able to find a drug that helps to limit nervous hunger and increase satiety, which unfortunately increases with the syndrome. Some moments are difficult to overcome. I hope to live as best as possible with the consequences of the syndrome, and I hope that doctors will find treatments to alleviate the risks. I hope that others can also live better.*
PWS caregivers	*– Surely his future will be full of satisfactions. We are working to ensure a positive future for him and a job that makes him happy and confident in his possibilities.* *– I believe the future will be difficult; I hope to be wrong, but I realised that most people just keep thinking about their needs. It is certainly difficult and challenging to care for or stay close to people with pathologies of this kind, but perhaps if everyone did their own and committed themselves more to the social, the future could be different. We should learn from an early age that we need to help others and be at the side of those in need.* *– As long as we, her family, are there, I think the future is "protected". The "after" is fearful, but I know I have to do something for her ... I owe her!*

DISCUSSION

The PRAXIS project represents the first effort to investigate PWS impact on HRQOL in Italy through NM, simultaneously considering the perspectives of underaged and adult patients with PWS and their caregivers. The project firstly aimed to understand their experience, real needs and personal resources. Fifty-five between children, adolescents, and adults with PWS reported joy and pride in sharing their stories, also suggesting that using evocative and open words[34] in structuring illness plots can be crucial to help people to express themselves. Moreover, the collection of 138 caregiving stories suggested a strong dedication to the survey and the need for caregivers to be listened to: they described writing as liberating, demonstrating its potentiality (a) to have a therapeutic

effect[27, 33] and (b) to be a safe room from the attitude of passing,[37] that is handling information considered discrediting or critical for the self to avoid social stigma.

Talking about PWS, indeed, still emerged as a "taboo". As shown in illness- and sickness-centred narratives [28], caregivers encountered significant difficulties in socialising the challenges PWS imposes in daily life, as well as the pain of having a child "different" from social imaginary: we may identify this as a social pain, also concerning caregivers in carrying on familiar criticalities. Furthermore, caregivers considered the project as a chance to invite society to integrate people with PWS and to denounce the stigma[37] still surrounding them.

If literature shows the cognitive impairment of people with PWS,[6] we would like to enrich evidences by suggesting to consider the multiple intelligences[38] these people show in their everyday experience. In line with Gardner's[38] reflection, revealing alternatives to the standard forms of intelligence (the logical-mathematical and linguistic ones), narratives showed the constant use of visuo-spatial, musical, interpersonal, existential, and introspective talents, resources and capabilities. In this regard, it has been reported that PWS patients show above the average performance in several tasks implying visuo-spatial skills,[39] which are linked with higher math abilities.[40] In particular, the importance of multiple intelligences emerged in food- control strategies and activities, consequently suggesting that considering them may positively influence overall HRQOL.

Considering the second purpose of the project, specific elements emerged from the analysis of therapeutic paths. PWS diagnosis mostly occurred up to the child's second year of life, but in some cases, a significant delay is still present, particularly for those people living away from specialised centres. Since timing is essential in PWS treatment,[9] training for neonatologists, geneticists and general paediatricians on PWS might improve early diagnosis. Moreover, other professionals

involved also need to develop or strengthen specific competencies on PWS to be able to address this condition.

Narratives showed some peculiar clinical PWS characteristics, such as irritability, aggressive and obsessive-compulsive[10, 11] and food-seeking[13] behaviours. The last resulted in the most challenging for both people with PWS and their caregivers: food management strategies,[41] as well as indoor and outdoor activities[42] and school or work schedules,[43, 44] can help people with PWS and caregivers to improve the relationship with food and within the family, consequently enhancing overall HRQOL.

Diet management and strategies, early GH therapy, clinical and psychiatric treatments, activities in specialised or social centres, and the different professional roles involved showed that a multi-professional approach integrating hospital and territory is fundamental to ensure adequate treatment of PWS,[17, 43, 45] as well as to mitigate the burden of caregiving reported both in narratives and literature.[14-16] In particular, two related topics emerged: (a) mostly women (mothers) changed or retired from work to caregiving; (b) family caregivers stated to be concerned on what will happen to their sons and daughters when they will remain alone – an issue already addressed by the Italian Law 112/2016 on social inclusion and autonomy of people with disabilities. These considerations also suggest, on the one hand, that social centres and services are crucial but need to be implemented in areas where lacking to support people with PWS and their caregivers, and on the other hand, that focusing on work policies is necessary to create autonomy and social inclusion.

The acknowledgement of the importance of multiple intelligences[38] in everyday experience also may improve HRQOL both of people with PWS or caregivers, together with helping in fighting the social stigma around PWS and enhancing social inclusion. Moreover, multiple intelligences might become a tool in clinical practice to evaluate people with PWS better and globally.

The participants in the project did not equally represent Italian different geographical areas, due to the local distribution of expert centres in the management of care for PWS, and this could be a selection bias. As for people with PWS, the inclusion criterium of being able to write represented a second bias.

In conclusion, this NM project allowed to gather new insights to PWS individual and social experiences, providing elements for improving a multidisciplinary approach to this condition: PWS social, relational, and emotional aspects crucially influence HRQOL and narratives can foster the relationship with these families.

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Competing interests

Authors have nothing to disclose.

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Author contributions

Conceptualisation: Ragusa, Crinò, Grugni, Reale, Marini. Analysis: Reale, Fiorencis. Investigation: Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Methodology: Reale, Marini. Project administration: Reale. Ragusa, Crinò, Grugni, Reale, Marini. Report visualisation: Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Methodology: Reale, Marini. Writing: Fiorencis, Ragusa, Crinò, Grugni, Reale, Marini. Editing: Fiorencis.

Data sharing

All data relevant to the project are included in the present manuscript. Original narratives are available into Italian upon request at the email areasanita@istud.it.

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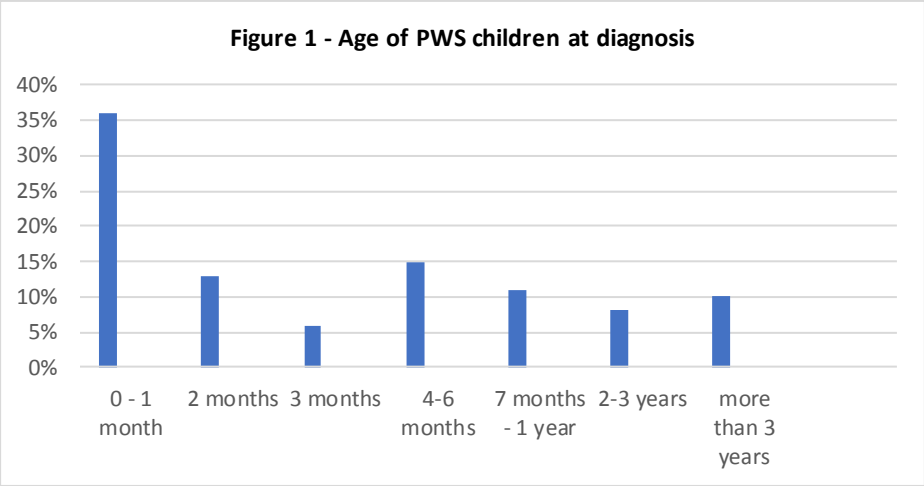
Figure legend

Figure 1 – Age of PWS children at diagnosis



Supplement 1

Medical centres for paediatric and adult patients of the Italian Network for Rare Disease

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Department of Paediatrics, Casa Sollievo della Sofferenza Research Institute, San Giovanni Rotondo (Foggia), Italy

Neonatology Unit, Spirito Santo Hospital, Pescara, Italy

Supplement 2

Illness plot addressed to underaged people with PWS

Hi! Can you tell us about yourself? […] May you introduce yourself and tell us about your life [...]. How do you feel with your friends […], your schoolmates […], and teachers? [...] You know that eating too much is not good for you: what do you do for not eating too much? [...] How do you feel with your family? […] Can you tell us about your visits to the doctors? […] What is your favorite game? […] What will you do when you grow up? [...]

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to adult people with PWS

I introduce myself [...]. I feel [...]; at home [...]; at work [...]. The people next to me [...]. The food [...]. The treatments [...]. What I like to do [...]. For me, I think of a future [...].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to caregivers of people with PWS

When I felt the first signs that something was going wrong […] I feel […]. They told us that it was […]. The hospitals I visited, the professionals I met […]. That period at home […], at work […]. The people next to me [...]. People around the person with PWS […]. Food […], and treatments […]. Today the person with PWS […], and treatments […]; I feel […]. At home, people with PWS […]. Outside domestic context, the person with PWS […]. What I like to do […]; what the person with PWS likes, beyond food […]. My life at home is […]; the food […]. I look at the person with PWS and I think for her/him a future […], and for me I imagine a future […].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Reporting checklist for qualitative study.

Based on the SRQR guidelines.

Instructions to authors

Complete this checklist by entering the page numbers from your manuscript where readers will find each of the items listed below.

Your article may not currently address all the items on the checklist. Please modify your text to include the missing information. If you are certain that an item does not apply, please write "n/a" and provide a short explanation.

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		Reporting Item	Page Number
Title			
	#1	Concise description of the nature and topic of the study identifying the study as qualitative or indicating the approach (e.g. ethnography, grounded theory) or data collection methods (e.g. interview, focus group) is recommended	1
Abstract			
	#2	Summary of the key elements of the study using the abstract format of the intended publication; typically includes background, purpose, methods, results and conclusions	1-2
Introduction			
Problem formulation	#3	Description and signifcance of the problem / phenomenon studied: review of relevant theory and empirical work; problem statement	2-4
Purpose or research question	#4	Purpose of the study and specific objectives or questions	4

Methods

Qualitative approach and research paradigm	#5	Qualitative approach (e.g. ethnography, grounded theory, case study, phenomenolgy, narrative research) and guiding theory if appropriate; identifying the research paradigm (e.g. postpositivist, constructivist / interpretivist) is also recommended; rationale. The rationale should briefly discuss the justification for choosing that theory, approach, method or technique rather than other options available; the assumptions and limitations implicit in those choices and how those choices influence study conclusions and transferability. As appropriate the rationale for several items might be discussed together.	4-6
Researcher characteristics and reflexivity	#6	Researchers' characteristics that may influence the research, including personal attributes, qualifications / experience, relationship with participants, assumptions and / or presuppositions; potential or actual interaction between researchers' characteristics and the research questions, approach, methods, results and / or transferability	n/a
Context	#7	Setting / site and salient contextual factors; rationale	4-5
Sampling strategy	#8	How and why research participants, documents, or events were selected; criteria for deciding when no further sampling was necessary (e.g. sampling saturation); rationale	5
Ethical issues pertaining to human subjects	#9	Documentation of approval by an appropriate ethics review board and participant consent, or explanation for lack thereof; other confidentiality and data security issues	5
Data collection methods	#10	Types of data collected; details of data collection procedures including (as appropriate) start and stop dates of data collection and analysis, iterative process, triangulation of sources / methods, and modification of procedures in response to evolving study findings; rationale	5-6
Data collection instruments and technologies	#11	Description of instruments (e.g. interview guides, questionnaires) and devices (e.g. audio recorders) used for data collection; if / how the instruments(s) changed over the course of the study	5-6

Units of study	#12	Number and relevant characteristics of participants, documents, or events included in the study; level of participation (could be reported in results)	6
Data processing	#13	Methods for processing data prior to and during analysis, including transcription, data entry, data management and security, verification of data integrity, data coding, and anonymisation / deidentification of excerpts	5-6
Data analysis	#14	Process by which inferences, themes, etc. were identified and developed, including the researchers involved in data analysis; usually references a specific paradigm or approach; rationale	6
Techniques to enhance trustworthiness	#15	Techniques to enhance trustworthiness and credibility of data analysis (e.g. member checking, audit trail, triangulation); rationale	5
Results/findings			
Syntheses and interpretation	#16	Main findings (e.g. interpretations, inferences, and themes); might include development of a theory or model, or integration with prior research or theory	6-8
Links to empirical data	#17	Evidence (e.g. quotes, field notes, text excerpts, photographs) to substantiate analytic findings	6-8
Discussion			
Intergration with prior work, implications, transferability and contribution(s) to the field	#18	Short summary of main findings; explanation of how findings and conclusions connect to, support, elaborate on, or challenge conclusions of earlier scholarship; discussion of scope of application / generalizability; identification of unique contributions(s) to scholarship in a discipline or field	8-11
Limitations	#19	Trustworthiness and limitations of findings	10
Other			
Conflicts of interest	#20	Potential sources of influence of perceived influence on study conduct and conclusions; how these were managed	1
Funding	#21	Sources of funding and other support; role of funders in data collection, interpretation and reporting	1

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BMJ Open

Caring and living with Prader–Willi syndrome in Italy: integrating children's, adults' and parents' experiences through a Narrative Medicine project

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Primary Subject Heading:	Qualitative research
Secondary Subject Heading:	Paediatrics, Diabetes and endocrinology
Keywords:	QUALITATIVE RESEARCH, Paediatric endocrinology < DIABETES & ENDOCRINOLOGY, Paediatric clinical genetics & dysmorphology < GENETICS, Eating disorders < PSYCHIATRY

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Caring and living with Prader–Willi syndrome in Italy: integrating children's, adults' and parents' experiences through a Narrative Medicine project

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ABSTRACT

Objectives. Prader–Willi syndrome (PWS) significantly impacts health-related quality of life (HRQoL); 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 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 underaged and adult patients with PWS and their caregivers. Written interviews, composed by a sociodemographic survey and a narrative, were collected through the project's website. Three dedicated illness plots employed evocative and open words to facilitate individual expression and to encourage reflection. Narratives were analysed through NVivo software. Researchers discussed the results with the project's steering committee.

Participants. Twenty-one children and adolescents and 34 adults with PWS joined the project, as well as 138 caregivers. A PWS diagnosis or the caregiving of a patient with PWS older than 5 years represented the eligibility criteria, as well as the willingness to share their illness experience by writing and the ability to communicate in Italian.

Results. The analysis of narratives led to understanding the PWS social and relational issues concerning diagnosis and current management, PWS daily experiences and social contexts, PWS implications in the working sphere, and participants' future perspectives. Narratives demonstrated that PWS management affects relationships and work-life balance and that social stigma remains present.

Conclusion. The project represented the first effort to investigate the impact of PWS on illness experience in Italy through NM while considering the perspectives of patients with PWS and their caregivers. The findings indicated that a multiprofessional approach is fundamental to ensure adequate treatment and provided elements for its improvement.

ARTICLE SUMMARY

Strengths and limitations of this study

- Inclusion of patients with PWS' perspective in the project
- Narrative medicine approach
- Participants did not equally represent the geographical areas of Italy
- Among patients with PWS, researchers included only those able to write

INTRODUCTION

Prader–Willi syndrome (PWS) is a rare genetic condition caused by an absence of functioning paternal genes on chromosome 15 in the 15q11-q13 region:[1] approximately 65%–70% of the cases are due to the deletion of this region, 20%–30% are caused by a maternal uniparental disomy (UPD) of chromosome 15, and most of the remaining 2%–5% have an imprinting centre defect or unbalanced translocations (~1%).[2] PWS occurs in approximately 1 in 10,000 to 30,000 births,[3] affecting both sexes and all geographic areas.[4]

Neonatal hypotonia, poor sucking and feeding difficulties characterise PWS in early infancy; dysmorphic signs (mild craniofacial abnormalities, small hands and feet, kyphoscoliosis), multiple endocrine abnormalities (growth hormone (GH)/IGF-I axis dysfunction, hypogonadism, central hypothyroidism, and central adrenal insufficiency) and developmental delay constitute other cardinal features of the syndrome.[5-7] Learning disabilities, maladaptive behaviours and hyperphagia—leading to life-threatening obesity if uncontrolled—follow in childhood and

adulthood.[3, 8] The mortality rate of patients with PWS is higher than in the general population,[9] with a 3% annual death rate across all ages.

Behavioural issues are noticeable in PWS, including aggressive and obsessive-compulsive behaviours and skin picking,[10, 11] and patients present a higher risk of developing psychiatric illness in adulthood;[12] food-seeking behaviours are particularly complex and [13] significantly affect patients' and caregivers' health-related quality of life (HRQoL). In particular, PWS caregivers—compared with other families managing children's disability or complex condition—report a higher level of stress, more difficulties in coping with symptoms,[14] a higher caregiving burden,[15] and a lower HRQoL.[16]

The clinical picture of patients with PWS substantially differs during the life span,[3, 6] and the prognosis is significantly conditioned by proactive interventions to prevent morbid weight excess.[9] Currently, no treatment is available for PWS. However, early diagnosis combined with multidisciplinary care favourably influences the course of PWS:[17] consequently, the diagnosis should be confirmed early during the neonatal period,[18] also with the support of genetic testing development.[19] In this context, early GH treatment has beneficial outcomes on, for example, height, body composition, endurance, and sense of well-being;[20-22] furthermore, early treatment with recombinant GH positively affects PWS[23] patients' and caregivers' HRQoL.[24, 25]

The social, relational, emotional, and existential aspects of PWS remain profoundly unknown, and the debate within Italian clinical and social communities has been poor: The World Health Organisation has stressed the importance of researching the measurable dimensions of HRQoL and—more broadly—illness experiences in leading clinical and social practice and recommends using narrative research.[26]

The discipline of narrative medicine (NM), based on illness narratives,[27] pursues the integration of the disease-centred approach and is concerned with clinical aspects and the illness-centred and sickness-centred approaches, which respectively focus on individual experience and the social understanding of a specific condition,[28] and both have often been neglected by the scientific community. The range of applications for narrative medicine is from clinical practice to therapeutic path design, education and research.[29] In research, narratives have demonstrated possible interventions on a specific condition through the integration of all perspectives involved in the pathway of care.[30, 31] Combining evidence-based medicine and NM provides clinicians methods to strengthen clinical practices with narrative competences.[29] NM research addresses the individual's experience when coping with distress caused by clinical conditions: It allows for the understanding of the profound experiences, needs and values of all actors involved in the care pathway.[27, 32] Scientific societies, healthcare facilities, and patient associations have increasingly employed NM research findings to improve the organisation and efficacy of healthcare services, generating sustainability[26] and fostering quality of care for patients and their social and relational contexts.[27]

The NM project "PRAXIS: Prader–Willi Excellence in Care with Story Taking" aimed to investigate the PWS illness experience by employing the analysis of narratives (a) to understand daily life, real needs and personal resources of people with PWS and their caregivers from diagnosis to current management, and by doing so, (b) furnish insights to support a multidisciplinary and a multiprofessional perspective in PWS clinical practice.[17] According to our review of the literature, no other project has addressed these issues simultaneously by considering the perspectives of underaged and adult patients with PWS and their caregivers.

METHODS

Patient and public involvement

The project was conducted in Italy between October 2018 and July 2019, as a part of a broader research project, and targeted people with PWS and their caregivers, as well as professionals working with PWS. The professionals underwent a webinar conducted by researchers from the "Istituto Studi Direzionali" (Institute of Management Studies, ISTUD) Foundation to be trained in NM and on the project's aims and methods; moreover, a parallel chart[27, 33] was identified as the most suitable NM tool to collect their narratives because it constitutes a personal notebook, parallel to the clinical record, in which professionals can write their impressions and feelings in plain language as a supplement to technical and quantitative reports.[27, 30, 33] Participants with PWS were given the possibility to express by drawing if under 5 years old or if unable to write; however, some participants over the threshold of 5 years old decided to maintain both the opportunities of expression.

The target group was people with PWS aged older than 5 years and their caregivers. Participants were recruited from 10 medical centres for paediatric and adult patients in the Italian Network for Rare Diseases (Supplement 1), namely six general hospitals and four scientific institutes of research, hospitalisation and healthcare: All the medical centres were macroregional, hospital-based centres that specialised in PWS treatment, and they were distributed among geographical areas (North, Central and South Italy). The Italian Prader–Willi Federation, and the Prader–Willi Association of the Lazio Region were also involved in disseminating the project; in particular, they organised three seminars—one each in the Lombardy, Lazio and Sicily regions—to provide the caregivers of those regions the opportunity to be further informed on NM and the project's aims and methods.

A PWS diagnosis, determined at the reference medical centre, or a caregiver for a person older than 5 years with PWS represented the eligibility criteria, as well as the willingness to share their illness experience by writing; thus, the ability to communicate in Italian was indispensable for the

inclusion in the project. Participants were informed of the possibility to view the projects (in Italian) on the project's webpage: www.medicinanarrativa.eu/praxis.

Data collection

Written stories of experiences were collected anonymously through the project's webpage; next, raw and anonymous narratives were downloaded as a Microsoft Excel spreadsheet. A sociodemographic survey constituted the written narrative, together with an illness plot[34, 35], namely, a plot related to the illness experience: It serves to guide narratives in a chronological order to identify evolutions over time and is characterised by evocative and open words that facilitate individual expression.[36]

Three illness plots were designed for three different groups—underage and adult patients with PWS, and caregivers (Supplement 2)—while addressing common aspects: (a) diagnosis and current management of the condition, namely the strategies related to food behaviours; (b) daily living with PWS, namely, the relational sphere and social context; and (c) the work experience and future perspectives.

The project design and the research tools were created by the project's steering committee, which comprised three endocrinologist experts in PWS, namely, one each from the Oasi Maria SS. Research Institute (Troina, Italy), the Bambino Gesù Paediatric Hospital of Palidoro (Rome, Italy), and the Istituto Auxologico Italiano of Piancavallo (Oggebbio, Italy), and three researchers from the ISTUD Foundation different for academic backgrounds, to reduce the personal influence on the research.

Ethical considerations

The project was performed according to the principles of the Declaration of Helsinki. Before the participants' involvement, they provided written informed consent after being briefed on the project's purpose and confidential data handling procedures, according to the Italian Law

196/2003 on Privacy and the Safeguarding of Sensitive Data.[37] Involved professionals obtained written informed consent to participate from parents and tutors for underaged participants during the first interview on the project's methods and purposes. Next, the professionals briefed the underage patients on the project and obtained their verbal consent to participate. Moreover, written informed consent to participate was obtained from adult participants and/or their tutors when appropriate. The Ethical Committee of the Oasi Maria SS. Research Institute (Troina, Italy) approved the project in January 2019 with the ethics approval number 2019/01/09/CE-IRCCS-OASI/19.

Analysis

We analysed the sociodemographic survey through descriptive statistics; no question was mandatory.

Anonymous narratives were entered into NVivo software[38] for coding and analysis. ISTUD researchers collectively coded 10 narratives in NVivo to assess consistency across team members. Afterwards, each narrative was coded separately by at least two researchers and then reviewed during weekly meetings and peer-debriefings to reduce bias in the interpretation of texts. Open interpretive coding was employed to identify and analyse emerging topics; Kleinman's[28] classification was retrospectively applied to the analysis of narratives because the researchers considered it the most suitable to further reveal illness-related and sickness-related aspects in narratives, respectively concerning the personal and emotional experience of a condition and how is it perceived within society.

The analysis process and results were shared within the project's steering committee to collectively address emerged topics and interpretation of data. Researchers followed the Standards for Reporting Qualitative Research (SRQR) Reporting Guidelines.[39]

RESULTS

Sociodemographic aspects

Twenty-one children and adolescents and 34 adults with PWS participated in the project, as well as 138 caregivers. Table 1 summarises the sociodemographic data of these three groups; the representation includes nonresponses as a separate category.

Table 1 – Sociodemographic data of participants

	Minors with PWS (N = 21)	Adults with PWS (N = 34)	PWS caregivers (N = 138)
Gender			
Females	6 (29%)	19 (56%)	99 (72%)
Males	15 (71%)	15 (44%)	37 (27%)
Nonresponses	0	0	2 (1%)
Age			
Average age (yrs)	14 (±3,09)	29(±9,72)	48(±9,04)
Minimum	7	19	20
Maximum	18	48	61
Geographic residence			
Northern Italy	3 (13%)	19 (58%)	29 (21%)
Central Italy	4 (19%)	6 (18%)	40 (29%)
Southern Italy	14 (69%)	9 (24%)	69 (50%)
Nonresponses	0	0	0
Education			
Elementary school	10 (48%)	2 (6%)	5 (4%)
Middle school	5 (24%)	10 (30%)	19 (14%)
High school	2 (10%)	21 (64%)	76 (55%)
University degree	0	1 (1%)	19 (14%)
Nonresponses	4 (19%)	0	19 (13%)
Marital status			
Single	21 (100%)	33 (97%)	3 (2%)
Married/ cohabitate	0	1 (3%)	118 (86%)
Divorced/separated	0	0	14 (10%)
Widowed	0	0	3 (2%)
Nonresponses	0	0	0
Employment status			
Student	21 (100%)	4 (11%)	3 (2%)
Working	0	19 (56%)	94 (68%)
Not working	0	11 (33%)	38 (28%)
Retired	0	0	3 (2%)
Nonresponses	0	0	0

Data presented as N (%) or mean (standard deviation) and minimum/maximum.

Results from the analysis are presented by following the dedicated illness plots' structure: (a) the first section concerns PWS diagnosis and current management, in which narratives' illness-related and sickness-related aspects, caregivers' perspectives on therapeutic path, and strategies to manage food-seeking behaviours are addressed; (b) the second section focuses on living with PWS

in relational and social contexts and addresses participants' indoor and outdoor daily activities; (c) the third section concerns caregivers' and PWS adult patients' narratives on the working sphere and participants' future perspectives and desires.

From diagnosis to the current management of PWS

Thirty-six percent of caregivers reported that their children were diagnosed with PWS within the first month of life (Figure 1); however, 10% affirmed that the diagnosis occurred after the child's third year of life.

In narratives, 95% of caregivers focused on PWS illness- and sickness-related aspects (Table 2); the remaining 5% adopted technical and clinical language[28] to discuss the condition, as exemplified in the following two quotes from narratives: (a) *She was hospitalised at the Neurology department for a muscle biopsy, she was diagnosed with congenital myopathy, and two years later, she underwent the DNA test; she was sent to the hospital in Northern Italy, and from there, we got the Prader–Willi diagnosis*; (b) *Adenoids, laryngotomy, broken arm fracture, desmoid, flat foot, strabismus, he has been taking GH since he was a child*. Disbelief, displacement, anger, and pain represented the most recurrent emotions expressed by caregivers when attempting to adapt to the situation and its criticalities. Patients with PWS—underaged and adult—described the condition only through its illness-related and sickness-related facets.

Table 2 – Illness- and sickness-related aspects: quotes from narratives

	Caregivers	Minors and adults with PWS
Illness	*– It is not easy to live with a child with this syndrome, especially when she asks for food. I tell her "No", and she starts crying and getting upset, screaming and telling me things I do not understand, but I do not give up.* *– Broken dreams, the feeling that something had changed forever, that my life had changed. I used to be so self-confident, so independent, and suddenly I felt fragile, scared and alone, dreadfully alone and unable to react.* *– The first emotion was fear. I did not know how to manage his crises for food. I did not understand what was happening to us, and every time we go to the doctor, we were reopening the wound, with all its fears.*	*– I feel proud, disability does not scare me.* *– Sometimes I feel happy, and sometimes sad because I have this disease.* *– I feel happy, sometimes a bit sad about my illness. But I am happy because the Lord created me, and I like myself as I came into existence.* *– I feel a little sad for having this rare disease; there are many difficulties, especially not overeating.*

	– When my son was born, doctors immediately told me that something was wrong. It was terrible. I was young, and he was my long-awaited first child.	
Sickness	*– Concern intensifies if I think that he will start school in September. I am worried about his level of tolerance if he will become the object of insults and bullying.* *– Taking care of a boy with Prader-Willi syndrome means changing the lifestyles of the whole family and combining the needs related to the disease with those of the family. It is a different journey, which could limit life experiences to those who do not have this kind of availability.* *– If I think of my child and her syndrome, I feel a great sorrow because I do not know if she will ever have a normal life.* *– Often, we felt desperate, especially thinking about our son's future. Then we learned to deal with problems as they arose.*	*– Other people are bad. They do not care about me because I have the syndrome.* *– I do not feel up to the kids of my generation. I hate me, I want to die. Sometimes I would like to be a boy like the others, and always be happy.* *– Prader-Willi syndrome lasts a lifetime. It is difficult to communicate with others, even with parents in the family.* *– On the one hand, I feel different from others because I am disabled and hypotonic. On the other hand, I feel like them because I was lucky enough to walk, to see and to hear.*

In Table 3, a focus on therapeutic paths from the caregivers' perspective meant addressing (a) relationships with professionals and (b) healthcare structures, both differing for specialisation and competence in PWS, and the (c) necessary or employed treatments, beyond diet.

Table 3 – PWS caregivers' perspective on therapeutic path: quotes from narratives

Professionals	*– Thanks to the specialist, we set up an appointment at the hospital, where we are currently followed. The cases they deal with are many, and for us, this is a guarantee. On a human level, they are unparalleled, available, smiling, and this allows you to spend more peaceful time, without feeling a burden.* *– I started attending health workers, support teachers and incapable teachers, unprepared paediatricians, arrogant doctors, medical commissions, courts. I clashed with bureaucracy, absurd health protocols, illogical rules to get what my daughter was entitled to and her psycho-physical well-being.* *– I was disappointed by the professionals. They talked about my son and my life as something unmodifiable, fatal, hopeless. I decided to get as far away as possible. My son was not a syndrome, he was a child.*
Healthcare structures	*– We do not live in a big city, so we had great difficulty getting our daughter adequately followed.* *– At the hospital they showed us the path we should have taken... on a medical level! On a therapeutic level, I had to resort to private professionals and centres.* *– Miles of road and hotel rooms characterise the therapeutic path that weighs on the family, both in economic terms and stress.*
Treatments	*– This part turned out to be better than expected. The only medicine is GH, which is simple and painless.* *– Psychomotricity, GH, speech and music therapy, psychological support and sport.* *– During adolescence, we had to resort to psychiatric drugs: this was a defeat for us, but it was necessary, our son had become unmanageable. Psychiatrists have a little knowledge of the syndrome.*

From caregivers' narratives, food-seeking behaviours[13] emerged as the most challenging event within the domestic context. Caregivers were aware that feeding is the first treatment for people with PWS and sought strategies to feed them. Fifty percent declared that they had achieved a balance, and the other 50% reported a problematic relationship with food. Both underaged and adult participants with PWS were aware of the importance of following a diet: positive or negative relationships with food emerged from narratives, where (a) the positive relationships also represented the result of commitment and several strategies to manage food-seeking behaviours, and (b) food-seeking was related to emotions (e.g. anger). Table 4 shows the main elements that emerged from the caregivers' and PWS participants' narratives.

Table 4 – Attitudes towards food-seeking behaviours: quotes from narratives

	Caregivers	Minors and adults with PWS
Commitment	*– The first thing I did was going to visit a professor in Switzerland to find a diet as clean as possible that could give strength and good energy to my child. I decided to follow it too.* *– We started a different relationship with food, and to be more aware of what we could and could not eat.* *– We tried to get the whole family accustomed to healthy eating as much as possible. I teach my son to read nutrition labels so he can choose the best one day. He already knows that he must not overeat sugar and fat; we eat at precise times, but managing hunger is not easy.*	*– I have a good relationship with food, I can control myself, I eat legumes and soups, and pasta in the evening. I really like vegetables.* *– I follow the correct diet, and I never steal food. If someone offers me something, I can refuse.* *– I am following a diet prepared by a nutritionist.*
Strategies	*– We have almost always managed to keep it under control; we focused a lot on food education (salads, vegetables, no snacks). We gave her the possibility to choose among certain foods. Therefore, she was more satisfied, allowing us to eat differently.* *– Food is the main problem for my child; there are both positive and negative moments. So, we are tough if we need to be tough, but sometimes we make some exceptions.* *– Food is always on our minds, but we try to manage everything at best. We try to live a life as normal as possible, and we try not to upset his habits. We give him some extras (he goes to parties, out with some friends, and to eat pizza).*	*– I like to eat everything. I try to eat lots of vegetables, even if this does not fill me, and whole wheat pasta. When I play or do a puzzle, I do not think about hunger. Mom tells me that if I want to eat more, I have to do more movement.* *– I try to organise my day according to fixed patterns, and I know I can eat at certain times.* *– In order not to think about food, I go for long walks, I do crossword puzzles and puzzles, I play tablet and computer.*
Criticalities	*– Unfortunately, food is an obsession and challenging to manage. Many times, I found my little child with hidden food.* *– The food issue becomes a daily challenge. Lunches and dinners are no longer quiet, and*	*– I eat out of meal because I hate me… I am hungry because my parents do not give me the right portions, and then I get fat and hospitalised, I steal food and go to the bar because I am hungry. I do not follow the diet.*

	you live very anxiously. The kitchen is no longer a meeting place. *– It is our conviction, a continuous struggle, day and night, the monster we have to defeat.*	*– Seeing the other people eating is painful.* *– When I think of food, my eyes shine, and when I see something to eat, I want it at all costs, and I cannot stop myself. I am always hungry, and I never get enough; when I overeat, I feel sick. When they tell me that I cannot eat so much, I get angry and nervous.*

Living with PWS in relationships and in social contexts

Thirty-six percent of PWS caregivers described daily life at home as quiet; however, most (64%) reported: fatigue (21%), chaos (6%), all-encompassing assistance (20%), and using tested routines to better manage food-seeking behaviours (17%). They have attempted to maintain their hobbies, interests and outside activities, even though their sons and daughters have PWS (Table 5). Relationships external to the family are difficult to preserve, imposing a radical change in social life. Indoor and outdoor activities represented an essential tool for caregivers in managing emotion patterns and food-seeking behaviours: narratives demonstrated that underaged and adult participants with PWS were aware of that. Sport, mind activity games, gardening and pet therapy were some of the most helpful activities reported; furthermore, patients with PWS appeared dedicated to cleanliness and routine activities. Both relational and activity spheres revealed the influence of behavioural and emotional changes in daily life and in familiar and social contexts; moreover, narratives addressed the strong presence of caregivers, as well as situations of social inclusion or exclusion.

Table 5 – Living with PWS in activities and relationships: quotes from narratives

	Caregivers	Minors and adults with PWS
Indoor activities	*– I like talking to my daughter, finding out what she wants to do and meeting her demands. I like to see her play with her friends and laugh because she is lovely and charismatic.* *– I love being with my family; I'm a full-time mom. I would like to go to the gym and to be a woman like the others, but I do not have time.* *– In my spare time I am a musician.*	*– I like reading fairy tales and colouring.* *– Mom does not want me to go to the kitchen; I do puzzles or embroidery; I see Walt Disney cartoons. I read the newspapers. When I am sad or angry, I try to eat. Often, I quarrel with my mom. My dad says nothing because he makes me feel more upset.* *– I see the television, I hear the music, and I pray every day. Every morning I make the beds, and I lay the table while my parents are cooking.*
Outdoor activities	*– I reload by being alone. I do yoga, I study Japanese, I sing in a choir. I do not give up my*	*– I play sports, especially football and swimming. I go for walks with a company,*

	passions. *– I like spending time with my family. I cultivate my hobbies, thanks to my wife's patience. With her, I share a love for concerts and cinema. I also cultivate my personal passions: wine and stickers collection.* *– I would like to swim, to go for walks by the sea and the lake. I would like to have no responsibilities, schedules, limitations; I would like to be a little carefree.*	*alone or with my dog. I like going to see my favourite football team at the stadium. I play videogames and watch movies. I like going to see my favourite animal, the dolphin, at the dolphinarium.* *– I have so many hobbies: dancing, singing, painting. I like art and going to the cinema and to the theater. I like reading crime novels, and I really like being with other people and playing cards, when the people around me do not fool me.* *– I like going out, shopping, swimming, listening to music, drawing, cutting and pasting, sticking figurines, making decoupage.*
Relationships	*– Some people have tried to understand the condition, others have disappeared. In this case, the syndrome helped me to make a selection among the people around me.* *– My father and my mother were as sorry and incredulous as I was, unable to get over it. My sister and friends supported me. My husband has shut himself off, present and absent at the same time, except for practical things.* *– Grandparents have been fantastic, available, always ready to help us. Friends did not understand our situation, and our roads soon split up: social life changes radically.*	*– My parents are the closest and most valuable people. They are strict but affectionate and loving. I am morbid with them, and I always look for contact even if I make them angry. My brothers are also close to me. One is playful, and the other is protective, a little severe. With them, I am serene and protective.* *– At school, I try to get closer to the other people, but they run away, they do not want me. Professors love me, even if with some of them, I feel I am different and not good. My parents: my biological mother did not love me, and my father hated me; my other parents love me so much.* *– The people next to me are my parents and my sister, and my personal support teachers. I do not like all my educators and my classmates because they spite me, but some of them are good.*

Work and future perspectives

Sixty-two percent of family caregivers had to change their job after the birth of their child with PWS (Table 6): more than one third left their current work, 8% changed jobs to assist the child, 8% requested a part-time job, and 3% abandoned the perspective of a career. From a gender perspective, 63% of female (mothers) and 33% of male caregivers had to change their current job to adapt to the child's condition. Forty-six percent did not discuss PWS in the workplace or discussed PWS with only their closest colleagues; 36% reported comprehensive behaviours, and 18% declared a lower understanding of PWS management necessities than for other diseases, such as cancer. Based on the narratives, work was a positive personal resource. Fifty-six percent of adults with PWS declared that they were working in jobs mainly characterised by low complexity

and repetitive operations and conducted in social cooperatives or centres, small companies with high corporate social responsibility, and family companies. Work is a source of pride and well-being and a distraction from food, but episodes of irritability and aggressive behaviours have been reported.

Table 6 – PWS and the working sphere: quotes from narratives

Adults with PWS	*– I am happy because my colleagues are great and they care about me, they are very close to me. I am glad because I like my job.* *– I deal with the press review, I answer by phone, I receive the people who come, and I do other tasks that the office managers assign me. If I am happy and motivated, I do not go looking for food. I look for food only when I am nervous. Now I have made so much progress, and therefore I do it less.* *– I often have nervous attacks, and I answer badly, offending with swear words those who make fun of me on how I do my job.* *– I work in a nursing home five days a week. I wash laundry, iron and fold the clothes of the guests. I work with a girl who follows me and teaches me many things. Every day I work with a different person, it depends on the shifts. With them, I feel good because they treat me like a normal person, and they congratulate me every time I work with them.*
PWS caregivers	*– My colleagues firmly supported me. They gave me the priority for any personal needs, so to allow me to not leave my son and my wife alone. They were like a second family to me, they all contributed as much as they could.* *– I had to leave work. I was working in the factory eight hours a day, and it was not possible to ask for a part-time job.* *– The job was a safety valve, a place to have a normal life.* *– At work, at first, there were no changes, because I did not immediately say what was happening. I talked about it with a few trusted colleagues, who showed me great warmth and solidarity.*

Regarding future perspectives, PWS family caregivers hope to have long lives so that they can care for their sons and daughters as long as possible, and they were particularly concerned with what would happen to their children without familial support (Table 7). Caregivers also addressed social inclusion, such as social changes and openness, rather than clinical solutions to PWS. Adult participants with PWS demonstrated self-realisation through work (27%)—as underaged participants did—and the desire to have a family (46%), recover from PWS (10%), and generally live well (14%).

Table 7 – Living with PWS and future perspectives: quotes from narratives

Minors with PWS	*– When I grow up, I want to be a professor of endocrinology to study my syndrome.* *– I want to be a vet because I love animals.* *– I want to drive an ambulance and do a lot of jobs.*
Adults with PWS	*– I want to start a family by adopting a child because I have so much love to give and being with my boyfriend. I hope to work in the same place where I currently am. I do*

	not know how to live without my parents, they help us a lot. *– I want to go living with the person I love the most in the world and to have children. Above all, I want to become a famous singer, and personally meet all my favourite singers. I want to get married and go to live in a beautiful house with a pool.* *– I hope that medicine will be able to find a drug to increase satiety and to limit nervous hunger, which is unfortunately increased by the syndrome. Some moments are hard to overcome. I hope to live as well as I can with the consequences of the syndrome, and I hope that doctors will find treatments to alleviate the risks. I hope that other people can also live better.*
PWS caregivers	*– I am sure that his future will be full of satisfactions. We are working to ensure him a positive future and a job that makes him happy and confident in his possibilities.* *– I believe the future will be challenging; I hope to be wrong, but I realised that most people mind their own business. Indeed, it is painful and challenging to care for or stay close to people with this kind of pathologies. Still, perhaps if people did their part and committed themselves more to the social, the future could be different. We should learn from an early age that we need to help other people and stand by those in need.* *– As long as we, her family, are there, I think the future is "protected". The "after" is fearful, but I know I have to do something for her ... I owe her!*

DISCUSSION

The PRAXIS project represents the first effort to investigate the PWS illness experience in Italy through NM by simultaneously considering the perspectives of underaged and adult patients with PWS and their caregivers. The project first aimed to understand their daily life, real needs and personal resources. Fifty-five participants, namely, children, adolescents and adults with PWS, reported joy and pride in sharing their stories, also suggesting that using evocative and open words[36] in structuring illness plots can be crucial to helping people to express themselves. Moreover, the collection of 138 caregiving stories suggested a strong dedication to the survey and the need for caregivers to be listened to: They described writing as liberating, demonstrating its potential (a) to have a therapeutic effect[27, 33] and (b) to be a safe space from the attitude of passing,[40] namely, handling information considered discrediting or critical for the self to avoid social stigma.

Talking about PWS emerged as a 'taboo'. In illness-centred and sickness-centred narratives [28], caregivers encountered significant difficulties in socialising the challenges PWS imposes in daily life, as well as the pain of having a child "different" from social imagery: We identify this as a social pain that also concerns caregivers when performing familiar criticalities. Furthermore, caregivers

considered the project a chance to invite society to integrate people with PWS and to denounce the stigma[40] that surrounds them.

If the literature demonstrates cognitive impairment in people with PWS,[6] we would like to enrich the evidence by suggesting the consideration of the multiple intelligences[41] these people demonstrate in their everyday experience. In line with Gardner's[41] reflection, revealing alternatives to the standard forms of intelligence (the logical-mathematical and linguistic ones), the narratives demonstrated the constant use of visuospatial, musical, interpersonal, existential and introspective talents, resources and capabilities. In this regard, patients with PWS have been reported to show above-average performance in several tasks implying visuospatial skills,[42] which in the general population are linked with higher math abilities.[43] In particular, the importance of multiple intelligences emerged in food- control strategies and activities, consequently suggesting that considering them may positively influence the overall illness experience.

Considering the second purpose of the project, specific elements emerged from the analysis of therapeutic paths. PWS diagnosis mostly occurred up to the child's second year of life, but in some cases, a significant delay remained, particularly for those people living away from specialised centres. Because timing is essential in PWS treatment,[9] training for neonatologists, geneticists and general paediatricians on PWS might improve early diagnosis. Moreover, the other professionals involved also must develop or strengthen specific PWS competencies to appropriately address this condition.

The narratives demonstrated some peculiar clinical PWS characteristics, such as irritability, aggressive and obsessive-compulsive behaviour [10, 11] and food-seeking[13] behaviours. The last resulted in the most challenges for the people with PWS and their caregivers: Food management strategies,[44] and indoor and outdoor activities[45] and school or work schedules,[46, 47] can

help people with PWS and their caregivers improve their relationships with food and family members, enhancing overall daily life.

Diet management and strategies, early GH therapy, clinical and psychiatric treatments, activities in specialised or social centres, and the different professional roles involved demonstrated that a mutiprofessional approach that integrates the factors of hospital and territory is fundamental to ensure adequate treatment of PWS [17, 46, 48] and to mitigate the burden of caregiving reported in the narratives and the literature.[14-16] In particular, two related topics emerged: (a) mostly women (mothers) changed or retired from work to become a caregiver; (b) family caregivers stated their concerns regarding what will happen to their sons and daughters if no family members are available —a topic already addressed by Italian Law 112/2016, on the social inclusion and autonomy of people with disabilities. These considerations also suggest, on the one hand, that social centres and services are crucial but need to be implemented in areas that have insufficient support for people with PWS and their caregivers, and on the other hand, a focus on work policies to create autonomy and social inclusion. Overcoming economic, legal, and social barriers and improving the current service provision still represent a challenge; patient organisations and scientific societies may have a crucial role in addressing these issues. Furthermore, although a National Plan for rare diseases[49] has been developed in Italy since 2013, its application in daily practice remains demanding. One possible intervention strategy to reduce the medical barriers requires universities and scientific societies to develop specific educational programs; in particular, creating a PWS national register may help to interface with similar international tools.[50]

The acknowledgement of the importance of multiple intelligences[41] in everyday experiences may also improve the daily and relational life of people with PWS or their caregivers, together with ameliorating the social stigma of PWS and enhancing social inclusion. Multiple intelligences

might also become a tool in clinical practice to better evaluate people with PWS; moreover, an evolving model for PWS care should include modern technologies, e.g. video visits, remote monitoring, and electronic health records.[51]

The participants in the project did not equally represent the different geographical areas in Italy because of the local distribution of expert centres in the management of care for PWS, and this could be a selection bias. Furthermore, the results are specific because of (a) the voluntary nature of the project and (b) the critical difference among Italian regional healthcare systems; therefore, further analysis is required. For people with PWS, the inclusion criterion of being able to write represented another bias.

In conclusion, this NM project provides new insights into the individual and social experiences related to PWS and provides elements for improving a multidisciplinary and multiprofessional perspectives on this condition: The social, relational, and emotional aspects of PWS crucially influence the illness experience and narratives that can foster the relationship between PWS professionals, patients, families and the community.

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Competing interests

Authors have nothing to disclose.

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Author contributions

Conceptualisation: Ragusa, Crinò, Grugni, Reale, Marini. Analysis: Reale, Fiorencis. Investigation: Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Methodology: Reale, Marini. Project administration: Reale. Ragusa, Crinò, Grugni, Reale, Marini. Report visualisation: Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Methodology: Reale, Marini. Writing: Fiorencis, Ragusa, Crinò, Grugni, Reale, Marini. Editing: Fiorencis.

Data sharing

All data relevant to the project are included in the present manuscript. Original narratives are available into Italian upon request at the email areasanita@istud.it.

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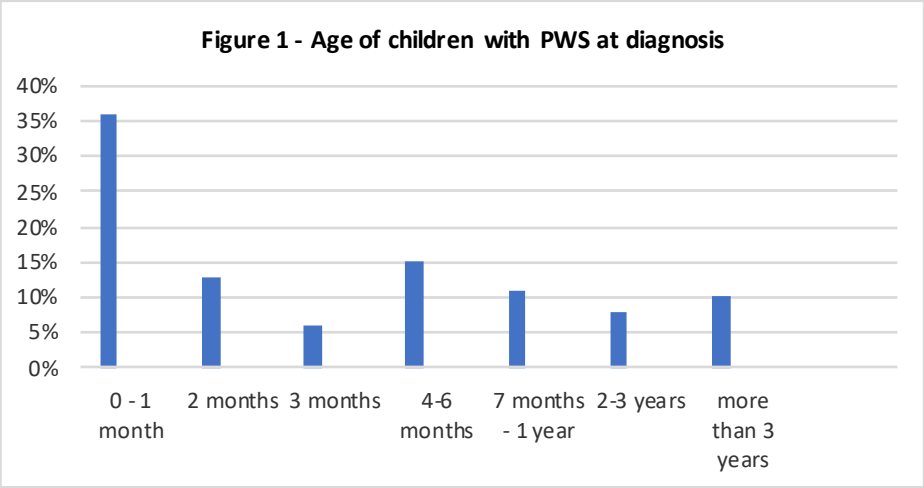
Figure legend

Figure 1 – Age of children with PWS at diagnosis



Supplement 1

Medical centres for paediatric and adult patients of the Italian Network for Rare Disease

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Bambino Gesù Paediatric Hospital, Research Institute, Palidoro (Rome), Italy

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Department of Biomedical Sciences and Human Oncology, University of Bari "A. Moro", Bari, Italy

Department of Human Pathology of Adulthood and Childhood "G.Barresi", University of Messina, Italy

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Department of Translational Sciences, University Federico II, Naples, Italy

Department of Paediatrics, Casa Sollievo della Sofferenza Research Institute, San Giovanni Rotondo (Foggia), Italy

Neonatology Unit, Spirito Santo Hospital, Pescara, Italy

Supplement 2

Illness plot addressed to underaged people with PWS

Hi! Can you tell us about yourself? […] May you introduce yourself and tell us about your life [...]. How do you feel with your friends […], your schoolmates […], and teachers? [...] You know that eating too much is not good for you: what do you do for not eating too much? [...] How do you feel with your family? […] Can you tell us about your visits to the doctors? […] What is your favorite game? […] What will you do when you grow up? [...]

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to adult people with PWS

I introduce myself [...]. I feel [...]; at home [...]; at work [...]. The people next to me [...]. The food [...]. The treatments [...]. What I like to do [...]. For me, I think of a future [...].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to caregivers of people with PWS

When I felt the first signs that something was going wrong […] I feel […]. They told us that it was […]. The hospitals I visited, the professionals I met […]. That period at home […], at work […]. The people next to me [...]. People around the person with PWS […]. Food […], and treatments […]. Today the person with PWS […], and treatments […]; I feel […]. At home, people with PWS […]. Outside domestic context, the person with PWS […]. What I like to do […]; what the person with PWS likes, beyond food […]. My life at home is […]; the food […]. I look at the person with PWS and I think for her/him a future […], and for me I imagine a future […].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Reporting checklist for qualitative study.

Based on the SRQR guidelines.

Instructions to authors

Complete this checklist by entering the page numbers from your manuscript where readers will find each of the items listed below.

Your article may not currently address all the items on the checklist. Please modify your text to include the missing information. If you are certain that an item does not apply, please write "n/a" and provide a short explanation.

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In your methods section, say that you used the SRQRreporting guidelines, and cite them as:

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		Reporting Item	Page Number
Title			
	#1	Concise description of the nature and topic of the study identifying the study as qualitative or indicating the approach (e.g. ethnography, grounded theory) or data collection methods (e.g. interview, focus group) is recommended	1
Abstract			
	#2	Summary of the key elements of the study using the abstract format of the intended publication; typically includes background, purpose, methods, results and conclusions	2
Introduction			
Problem formulation	#3	Description and signifcance of the problem / phenomenon studied: review of relevant theory and empirical work; problem statement	3-4
Purpose or research question	#4	Purpose of the study and specific objectives or questions	5

Methods			
Qualitative approach and research paradigm	#5	Qualitative approach (e.g. ethnography, grounded theory, case study, phenomenolgy, narrative research) and guiding theory if appropriate; identifying the research paradigm (e.g. postpositivist, constructivist / interpretivist) is also recommended; rationale. The rationale should briefly discuss the justification for choosing that theory, approach, method or technique rather than other options available; the assumptions and limitations implicit in those choices and how those choices influence study conclusions and transferability. As appropriate the rationale for several items might be discussed together.	4-6
Researcher characteristics and reflexivity	#6	Researchers' characteristics that may influence the research, including personal attributes, qualifications / experience, relationship with participants, assumptions and / or presuppositions; potential or actual interaction between researchers' characteristics and the research questions, approach, methods, results and / or transferability	7
Context	#7	Setting / site and salient contextual factors; rationale	5-6
Sampling strategy	#8	How and why research participants, documents, or events were selected; criteria for deciding when no further sampling was necessary (e.g. sampling saturation); rationale	6
Ethical issues pertaining to human subjects	#9	Documentation of approval by an appropriate ethics review board and participant consent, or explanation for lack thereof; other confidentiality and data security issues	7-8
Data collection methods	#10	Types of data collected; details of data collection procedures including (as appropriate) start and stop dates of data collection and analysis, iterative process, triangulation of sources / methods, and modification of procedures in response to evolving study findings; rationale	7
Data collection instruments and technologies	#11	Description of instruments (e.g. interview guides, questionnaires) and devices (e.g. audio recorders) used for data collection; if / how the instruments(s) changed over the course of the study	7

Units of study	#12	Number and relevant characteristics of participants, documents, or events included in the study; level of participation (could be reported in results)	9
Data processing	#13	Methods for processing data prior to and during analysis, including transcription, data entry, data management and security, verification of data integrity, data coding, and anonymisation / deidentification of excerpts	8
Data analysis	#14	Process by which inferences, themes, etc. were identified and developed, including the researchers involved in data analysis; usually references a specific paradigm or approach; rationale	8
Techniques to enhance trustworthiness	#15	Techniques to enhance trustworthiness and credibility of data analysis (e.g. member checking, audit trail, triangulation); rationale	8
Results/findings			
Syntheses and interpretation	#16	Main findings (e.g. interpretations, inferences, and themes); might include development of a theory or model, or integration with prior research or theory	9-10
Links to empirical data	#17	Evidence (e.g. quotes, field notes, text excerpts, photographs) to substantiate analytic findings	10-16
Discussion			
Intergration with prior work, implications, transferability and contribution(s) to the field	#18	Short summary of main findings; explanation of how findings and conclusions connect to, support, elaborate on, or challenge conclusions of earlier scholarship; discussion of scope of application / generalizability; identification of unique contributions(s) to scholarship in a discipline or field	16-19
Limitations	#19	Trustworthiness and limitations of findings	19
Other			
Conflicts of interest	#20	Potential sources of influence of perceived influence on study conduct and conclusions; how these were managed	19
Funding	#21	Sources of funding and other support; role of funders in data collection, interpretation and reporting	19

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BMJ Open

Caring and living with Prader-Willi syndrome in Italy: integrating children's, adults' and parents' experiences through a multicentre Narrative Medicine research.

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Primary Subject Heading:	Qualitative research
Secondary Subject Heading:	Paediatrics, Diabetes and endocrinology
Keywords:	QUALITATIVE RESEARCH, Paediatric endocrinology < DIABETES & ENDOCRINOLOGY, Paediatric clinical genetics & dysmorphology < GENETICS, Eating disorders < PSYCHIATRY

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Caring and living with Prader–Willi syndrome in Italy: integrating children's, 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 (HRQoL); 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 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 underaged and adult patients with PWS and their caregivers. Written interviews, composed by a sociodemographic survey and a narrative, were collected through the project's website. Three dedicated illness plots employed evocative and open words to facilitate individual expression and to encourage reflection. Narratives were analysed through NVivo software. Researchers discussed the results with the project's steering committee.

Participants. Twenty-one children and adolescents and 34 adults with PWS joined the project, as well as 138 caregivers. A PWS diagnosis or the caregiving of a patient with PWS older than 5 years represented the eligibility criteria, as well as the willingness to share their illness experience by writing and the ability to communicate in Italian.

Results. The analysis of narratives led to understanding the PWS social and relational issues concerning diagnosis and current management, PWS daily experiences and social contexts, PWS implications in the working sphere, and participants' future perspectives. Narratives demonstrated that PWS management affects relationships and work-life balance and that social stigma remains present.

Conclusion. The project represented the first effort to investigate the impact of PWS on illness experience in Italy through NM while considering the perspectives of patients with PWS and their caregivers. The findings indicated that a multiprofessional approach is fundamental to ensure adequate treatment and provided elements for its improvement.

ARTICLE SUMMARY

Strengths and limitations of this study

- Inclusion of patients with PWS' perspective in the project
- Narrative medicine approach
- Participants did not equally represent the geographical areas of Italy
- Among patients with PWS, researchers included only those able to write

INTRODUCTION

Prader–Willi syndrome (PWS) is a rare genetic condition caused by an absence of functioning paternal genes on chromosome 15 in the 15q11-q13 region:[1] approximately 65%–70% of the cases are due to the deletion of this region, 20%–30% are caused by a maternal uniparental disomy (UPD) of chromosome 15, and most of the remaining 2%–5% have an imprinting centre defect or unbalanced translocations (~1%).[2] PWS occurs in approximately 1 in 10,000 to 30,000 births,[3] affecting both sexes and all geographic areas.[4]

Neonatal hypotonia, poor sucking and feeding difficulties characterise PWS in early infancy; dysmorphic signs (mild craniofacial abnormalities, small hands and feet, kyphoscoliosis), multiple endocrine abnormalities (growth hormone (GH)/IGF-I axis dysfunction, hypogonadism, central hypothyroidism, and central adrenal insufficiency) and developmental delay constitute other cardinal features of the syndrome.[5-7] Learning disabilities, maladaptive behaviours and hyperphagia—leading to life-threatening obesity if uncontrolled—follow in childhood and

adulthood.[3, 8] The mortality rate of patients with PWS is higher than in the general population,[9] with a 3% annual death rate across all ages.

Behavioural issues are noticeable in PWS, including aggressive and obsessive-compulsive behaviours and skin picking,[10, 11] and patients present a higher risk of developing psychiatric illness in adulthood;[12] food-seeking behaviours are particularly complex and [13] significantly affect patients' and caregivers' health-related quality of life (HRQoL). In particular, PWS caregivers—compared with other families managing children's disability or complex condition—report a higher level of stress, more difficulties in coping with symptoms,[14] a higher caregiving burden,[15] and a lower HRQoL.[16]

The clinical picture of patients with PWS substantially differs during the life span,[3, 6] and the prognosis is significantly conditioned by proactive interventions to prevent morbid weight excess.[9] Currently, no treatment is available for PWS. However, early diagnosis combined with multidisciplinary care favourably influences the course of PWS;[17] therefore, the diagnosis should be confirmed early during the neonatal period,[18] with the support of genetic testing development.[19] In this context, early GH treatment has beneficial outcomes on, for example, height, body composition, endurance, and sense of well-being;[20-22] furthermore, early treatment with recombinant GH positively affects PWS[23] patients' and caregivers' HRQoL.[24, 25]

The social, relational, emotional, and existential aspects of PWS remain profoundly unknown, and the debate within Italian clinical and social communities has been poor: The World Health Organisation has stressed the importance of researching the measurable dimensions of HRQoL and—more broadly—illness experiences in leading clinical and social practice and recommends using narrative research.[26]

The discipline of narrative medicine (NM), based on illness narratives,[27] pursues the integration of the disease-centred approach and is concerned with clinical aspects and the illness-centred and sickness-centred approaches, which respectively focus on individual experience and the social understanding of a specific condition,[28] and both have often been neglected by the scientific community. The range of applications for NM is from clinical practice to therapeutic path design, education and research.[29] In research, narratives have demonstrated possible interventions on a specific condition through the integration of all perspectives involved in the pathway of care.[30, 31] Combining evidence-based medicine and NM provides clinicians methods to strengthen clinical practices with narrative competences.[29] NM research addresses the individual's experience when coping with distress caused by clinical conditions: It allows for the understanding of the profound experiences, needs and values of all actors involved in the care pathway.[27, 32] Scientific societies, healthcare facilities, and patient associations have increasingly employed NM research findings to improve the organisation and efficacy of healthcare services, generating sustainability[26] and fostering quality of care for patients and their social and relational contexts.[27]

The NM project "PRAXIS: Prader–Willi Excellence in Care with Story Taking" aimed to investigate the PWS illness experience by employing the analysis of narratives (a) to understand daily life, real needs and personal resources of people with PWS and their caregivers from diagnosis to current management, and by doing so, (b) furnish insights to support a multidisciplinary and a multiprofessional perspective in PWS clinical practice.[17] According to our review of the literature, no other project has addressed these issues simultaneously by considering the perspectives of underaged and adult patients with PWS and their caregivers.

METHODS

Research design and setting

The project was conducted in Italy between October 2018 and July 2019, as a part of a broader research project, and targeted people with PWS and their caregivers, as well as professionals working with PWS. The professionals underwent a webinar conducted by researchers from the "Istituto Studi Direzionali" (Institute of Management Studies, ISTUD) Foundation to be trained in NM and on the project's aims and methods; moreover, a parallel chart[27, 33] was identified as the most suitable NM tool to collect their narratives because it constitutes a personal notebook, parallel to the clinical record, in which professionals can write their impressions and feelings in plain language as a supplement to technical and quantitative reports.[27, 30, 33] Participants with PWS were given the possibility to express by drawing if under 5 years old or if unable to write; however, some participants over the threshold of 5 years old decided to maintain both the opportunities of expression.

The target group was people with PWS aged older than 5 years and their caregivers. Participants were recruited from 10 medical centres for paediatric and adult patients in the Italian Network for Rare Diseases (Supplement 1), namely six general hospitals and four scientific institutes of research, hospitalisation and healthcare: All the medical centres were macroregional, hospital-based centres that specialised in PWS treatment, and they were distributed among geographical areas (North, Central and South Italy). The Italian Prader–Willi Federation, and the Prader–Willi Association of the Lazio Region were also involved in disseminating the project; in particular, they organised three seminars—one each in the Lombardy, Lazio and Sicily regions—to provide the caregivers of those regions the opportunity to be further informed on NM and the project's aims and methods.

A PWS diagnosis, determined at the reference medical centre, or a caregiver for a person older than 5 years with PWS represented the eligibility criteria, as well as the willingness to share their illness experience by writing; thus, the ability to communicate in Italian was indispensable for the

inclusion in the project. Participants were informed of the possibility to view the projects (in Italian) on the project's webpage: www.medicinanarrativa.eu/praxis.

Data collection

Written stories of experiences were collected anonymously through the project's webpage; next, raw and anonymous narratives were downloaded as a Microsoft Excel spreadsheet. A sociodemographic survey constituted the written narrative, together with an illness plot[34, 35], namely, a plot related to the illness experience: It serves to guide narratives in a chronological order to identify evolutions over time and is characterised by evocative and open words that facilitate individual expression.[36]

Three illness plots were designed for three different groups—underage and adult patients with PWS, and caregivers (Supplement 2)—while addressing common aspects: (a) diagnosis and current management of the condition, namely the strategies related to food behaviours; (b) daily living with PWS, namely, the relational sphere and social context; and (c) the work experience and future perspectives.

The project design and the research tools were created by the project's steering committee, which comprised three endocrinologist experts in PWS, namely, one each from the Oasi Maria SS. Research Institute (Troina, Italy), the Bambino Gesù Paediatric Hospital of Palidoro (Rome, Italy), and the Istituto Auxologico Italiano of Piancavallo (Oggebbio, Italy), and three researchers from the ISTUD Foundation different for academic backgrounds, to reduce the personal influence on the research.

Patient and public involvement

The research was conducted without patient involvement. Patients did not participate in developing the research design and tools and were not engaged in the interpretation and

discussion of the results. Patients were not invited to contribute to the writing or editing of this document.

Ethical considerations

The project was performed according to the principles of the Declaration of Helsinki. Before the participants' involvement, they provided written informed consent after being briefed on the project's purpose and confidential data handling procedures, according to the Italian Law 196/2003 on Privacy and the Safeguarding of Sensitive Data[37] and the General Data Protection Regulation of the European Union 2016/679.[38] Involved professionals obtained written informed consent to participate from parents and tutors for underaged participants during the first interview on the project's methods and purposes. Next, the professionals briefed the underage patients on the project and obtained their verbal consent to participate. Moreover, written informed consent to participate was obtained from adult participants and/or their tutors when appropriate. The Ethical Committee of the Oasi Maria SS. Research Institute (Troina, Italy) approved the project in January 2019 with the ethics approval number 2019/01/09/CE-IRCCS-OASI/19.

Analysis

We analysed the sociodemographic survey through descriptive statistics; no question was mandatory.

We separately examined caregivers' and patients' perspectives. Anonymous narratives were entered into NVivo software[39] for coding and analysis. ISTUD researchers collectively coded 10 narratives in NVivo to assess consistency across team members. Afterwards, each narrative was coded separately by at least two researchers and then reviewed during weekly meetings and peer-debriefings to reduce bias in the interpretation of texts. Open interpretive coding was employed to identify and analyse emerging topics; Kleinman's[28] classification was retrospectively applied

to the analysis of narratives because the researchers considered it the most suitable to further reveal illness-related and sickness-related aspects in narratives, respectively concerning the personal and emotional experience of a condition and how is it perceived within society.

The analysis process and results were shared within the project's steering committee to collectively address emerged topics and interpretation of data. Researchers followed the Standards for Reporting Qualitative Research Reporting Guidelines.[40]

RESULTS

Sociodemographic aspects

Twenty-one children and adolescents and 34 adults with PWS participated in the project, as well as 138 caregivers. Table 1 summarises the sociodemographic data of these three groups; the representation includes nonresponses as a separate category.

Table 1 – Sociodemographic data of participants

	Minors with PWS (N = 21)	Adults with PWS (N = 34)	PWS caregivers (N = 138)
Gender			
Females	6 (29%)	19 (56%)	99 (72%)
Males	15 (71%)	15 (44%)	37 (27%)
Nonresponses	0	0	2 (1%)
Age (yrs.)			
Mean (SD)	14 (3,09)	29(9,72)	48(9,04)
Minimum	7	19	20
Maximum	18	48	61
Geographic residence			
Northern Italy	3 (13%)	19 (58%)	29 (21%)
Central Italy	4 (19%)	6 (18%)	40 (29%)
Southern Italy	14 (69%)	9 (24%)	69 (50%)
Nonresponses	0	0	0
Education			
Elementary school	10 (48%)	2 (6%)	5 (4%)
Middle school	5 (24%)	10 (30%)	19 (14%)
High school	2 (10%)	21 (64%)	76 (55%)
University degree	0	1 (1%)	19 (14%)
Nonresponses	4 (19%)	0	19 (13%)
Marital status			
Single	21 (100%)	33 (97%)	3 (2%)
Married/ cohabitate	0	1 (3%)	118 (86%)
Divorced/separated	0	0	14 (10%)
Widowed	0	0	3 (2%)
Nonresponses	0	0	0
Employment status			
Student	21 (100%)	4 (11%)	3 (2%)

Working	0	19 (56%)	94 (68%)
Not working	0	11 (33%)	38 (28%)
Retired	0	0	3 (2%)
Nonresponses	0	0	0

Data presented as N (%) or mean (standard deviation) and minimum/maximum.

Results from the analysis are presented by following the dedicated illness plots' structure: (a) the first section concerns PWS diagnosis and current management, in which narratives' illness-related and sickness-related aspects, caregivers' perspectives on therapeutic path, and strategies to manage food-seeking behaviours are addressed; (b) the second section focuses on living with PWS in relational and social contexts and addresses participants' indoor and outdoor daily activities; (c) the third section concerns caregivers' and PWS adult patients' narratives on the working sphere and participants' future perspectives and desires.

From diagnosis to the current management of PWS

Thirty-six percent of caregivers reported that their children were diagnosed with PWS within the first month of life (Figure 1); however, 10% affirmed that the diagnosis occurred after the child's third year of life.

In narratives, 95% of caregivers focused on PWS illness- and sickness-related aspects (Table 2); the remaining 5% adopted technical and clinical language[28] to discuss the condition, as exemplified in the following two quotes from narratives. (a) *She was hospitalised at the Neurology department for a muscle biopsy; diagnosed with congenital myopathy and 2 years later, underwent the DNA test. She was sent to a hospital in Northern Italy, and from there, we got the Prader–Willi diagnosis* (caregiver 015). (b) *He underwent nine surgeries: adenoids, laryngotomy, broken arm fracture, desmoid, flat foot, strabismus. He has been taking GH since he was a child* (caregiver 063). Disbelief, displacement, anger, and pain represented the most recurrent emotions expressed by caregivers when attempting to adapt to the situation and its criticalities. Patients with PWS—

underaged and adult—described the condition only through its illness-related and sickness-related facets.

Table 2 – Illness- and sickness-related aspects: quotes from narratives

	Caregivers	Minors and adults with Prader–Willi syndrome
Illness	*– It is not easy to live with a child who has this syndrome, especially when she asks for food. I tell her "No," and she starts to cry and get upset, screaming and saying things to me that I do not understand, but I do not give up.* (Caregiver 051) *– I was nervous. I didn't know what was happening. Yet in the delivery room, when I saw my baby, she was beautiful! What was the problem? When I talked to a geneticist the next day, I started crying.* (Caregiver 074) *– Broken dreams, the feeling that something has changed forever—that my life has changed. I used to be so self-confident, so independent, and suddenly, I felt fragile, scared, and alone—dreadfully alone, unable to react.* (Caregiver 031) *– When my son was born, the doctors immediately told me that something was wrong. It was terrible. I was young, and he was my long-awaited first child.* (Caregiver 019)	*– I feel proud. My disability does not scare me.* (Adult with PWS 030) *– Sometimes I feel happy, and sometimes I am sad because of my disease.* (Adult with PWS 005) *– I want to be with the other non-disabled kids—my only problem is eating.* (Minor with PWS 007) *– I mostly feel happy, but sometimes I am a bit sad about my illness. I am happy because the Lord created me, and I like myself as I am when I came into existence.* (Adult with PWS 009)
Sickness	*– Food is not the only danger: anything can happen to my son, he can be tricked or manipulated.* (Caregiver 062) *– When my daughter was diagnosed with Prader–Willi syndrome, I felt terrible, because I couldn't accept the syndrome and because I saw the other mothers with their children, happy and carefree, and I knew it wouldn't be like that for us. Over time, I learned to accept the situation. My daughter's disability does not mean not living—it means living differently.* (Caregiver 049) *– I was upset. It was hard to believe that my daughter could not have a future like all the other children.* (Caregiver 052) *– Often, we felt desperate, especially as we thought about our son's future, but then, we learned to deal with problems as they arose.* (Caregiver 027)	*– I do not feel equal to the kids of my generation. I hate myself. I want to die. Sometimes I would like to be a boy like the others and to always be happy.* (Adult with PWS 014) *– The effects of Prader–Willi syndrome last throughout one's lifetime. It is difficult to communicate with others, even with my own parents.* (Adult with PWS 016) *– Other people are bad. They do not care about me because I have this syndrome.* (Minor with PWS 007) *– On the one hand, I feel different from others because I am disabled and hypotonic. On the other hand, I do feel like them because I am lucky enough to walk, to see and to hear.* (Adult with PWS 032)

In Table 3, a focus on therapeutic paths from the caregivers' perspective meant addressing (a) relationships with different professionals and (b) healthcare structures, and the (c) necessary or employed treatments, beyond diet.

Table 3 –Perspectives from PWS caregivers on the therapeutic path: quotes from narratives

Professionals	*– Thanks to our specialist, we got an appointment at the hospital, where we currently get care. They deal with many cases, and for us, this is a guarantee. On a human level, they are unparalleled, available, smiling; this lets us be more at peace, without feeling our burden.* (Caregiver 038) *– I started meeting health workers, support teachers and incapable teachers, unprepared paediatricians, arrogant doctors, medical commissions, and courts. I clashed with bureaucracy, absurd health protocols and illogical rules to get what my daughter was entitled to and to support her psycho-physical well-being.* (Caregiver 031) *– I was disappointed by the professionals. They talked about my son and my life as something that could not be changed, fatal, hopeless. I decided to get as far away as possible. My son was not a syndrome, he was a child.* (Caregiver 011)
Healthcare structures	*– We do not live in a big city, so we had great difficulty getting our daughter adequately seen to.* (Caregiver 018) *– At the hospital, they presented us with the path we should have taken... on a medical level! For a therapeutic level, I had to resort to private professionals and centres.* (Caregiver 024) *– Miles of roads and hotel rooms characterise the therapeutic path that families must pass, both in economic terms and in terms of stress.* (Caregiver 066)
Treatments	*– This part was better than expected. The only medicine we use is GH, which is simple and painless.* (Caregiver 008) *– Psychomotricity, GH, speech and music therapy, psychological support and sports.* (Caregiver 010) *– During his adolescence, we had to resort to psychiatric drugs: this felt like a defeat, but it was necessary. Our son had become unmanageable. Psychiatrists have little knowledge of the syndrome.* (Caregiver 027)

From caregivers' narratives, food-seeking behaviours[13] emerged as the most challenging event within the domestic context. Caregivers were aware that feeding is the first treatment for people with PWS and sought strategies to feed them. Fifty percent declared that they had achieved a balance, and the other 50% reported a problematic relationship with food. Both underaged and adult participants with PWS were aware of the importance of following a diet: positive or negative relationships with food emerged from narratives, where (a) the positive relationships also represented the result of commitment and several strategies to manage food-seeking behaviours, and (b) food-seeking was related to emotions (e.g. anger). Table 4 shows the main elements that emerged from the caregivers' and PWS participants' narratives.

Table 4 – Attitudes towards food-seeking behaviours: quotes from narratives

	Caregivers	Minors and adults with Prader–Willi syndrome
Commitment	*– The first thing that I did was to visit a professor in Switzerland to get a diet as pure as possible that could give strength and good energy to my child. I decided to follow it.* (Caregiver 011) *– We began to have a different relationship*	*– I know eating a lot is bad for me. I follow my diet, so I don't get fat.* (Minor with PWS 005) *– I stay on my diet, and I never steal food. Even if someone offers me something, I do not accept it.* (Adult with PWS 032) *– I am following a diet that was prepared by a*

	with food and to be more aware of what we could and could not eat. (Caregiver 034) *– We tried to accustom the whole family to healthy eating as much as possible. I taught my son to read nutrition labels so that later he would be able to choose what is best. He already knows that he must not eat too much sugar and fat. We eat at fixed times, but it is not easy to manage his hunger.* (Caregiver 040)	*nutritionist.* (Adult with PWS 023)
Strategies	*– We are almost always able to keep it under control; we focus a lot on food education (i.e. salads, vegetables, and no snacks). We let her choose among certain foods. This helped satisfy her, allowing us to eat differently.* (Caregiver 032) *– Food is the main problem for my child; there are both positive and negative moments. So, we are tough if we need to be, but sometimes we make exceptions.* (Caregiver 060) *– Food is always on our minds, but we try to manage everything in the best way we can. We try to live as normal a life as possible, and we try not to upset his habits. We give him some treats (he goes to parties, goes out with some friends, and can eat pizza).* (Caregiver 064)	*– I like to eat everything. I try to eat lots of vegetables, even if I do not feel full, and whole wheat pasta. When I am playing or doing a puzzle, I do not think about hunger. Mom tells me that if I want to eat more, I have to move around more.* (Adult with PWS 003) *– I try to organise my day in fixed patterns, and I know I can eat at certain times.* (Minor with PWS 003) *– So, I do not think about food, I go for long walks, I do crossword puzzles and other puzzles and I play on my tablet and computer.* (Minor with PWS 006)
Criticalities	*– Unfortunately, food is an obsession and is challenging to manage. I have found my little child hiding food many times.* (Caregiver 052) *– The food issue is a daily challenge. Lunches and dinners are no longer quiet, and we live with anxiety. The kitchen is no longer a meeting place.* (Caregiver 066) *– This is our conviction, a continuous struggle, day and night—the monster we have to defeat.* (Caregiver 067)	*– I eat outside of meals because I hate myself… I am hungry because my parents do not give me the right portions, and then I get fat and go to the hospital, I steal food… because I am hungry. I do not follow my diet.* (Adult with PWS 014) *– Seeing other people eating is painful.* (Adult with PWS 022) *– When I think of food, my eyes shine, and when I see food, I want it at all costs, and I cannot stop myself. I am always hungry, and I never get enough; when I overeat, I feel sick. When they tell me that I should not eat so much, I get angry and anxious.* (Adult 031)

Living with PWS in relationships and in social contexts

Thirty-six percent of PWS caregivers described daily life at home as quiet; however, most (64%) reported: fatigue (21%), chaos (6%), all-encompassing assistance (20%), and using tested routines to better manage food-seeking behaviours (17%). They have attempted to maintain their hobbies, interests and outside activities, even though their sons and daughters have PWS (Table 5). Relationships external to the family are difficult to preserve, imposing a radical change in social life. Indoor and outdoor activities represented an essential tool for caregivers in managing

emotion patterns and food-seeking behaviours: narratives demonstrated that underaged and adult participants with PWS were aware of that. Sport, mind activity games, gardening and pet therapy were some of the most helpful activities reported; furthermore, patients with PWS appeared dedicated to cleanliness and routine activities. Both relational and activity spheres revealed the influence of behavioural and emotional changes in daily life and in familiar and social contexts; moreover, narratives addressed the strong presence of caregivers, as well as situations of social inclusion or exclusion.

Table 5 – Living with Prader–Willi syndrome (PWS) in activities and relationships: quotes from narratives

	Caregivers	Minors and adults with PWS
Indoor activities	*– I like talking to my daughter, finding out what she wants to do and meeting her needs. I like to see her play with her friends and laugh because she is lovely and charismatic.* (Caregiver 037) *– I love being with my family; I'm a full-time mom. I would like to go to the gym and to be a woman like others, but I do not have time.* (Caregiver 043) *– In my spare time I am a musician.* (Caregiver 041)	*– I have two kittens at home. I take care of them. At home, I help my mother cleaning and ironing clothes and shirts. Every day I take care of my hygiene and make my bed.* (Minor with PWS 002) *– Mom does not want me to go to the kitchen. I do puzzles or embroidery. I watch Disney cartoons. I read the newspapers. When I am sad or angry, I try to eat. I often quarrel with my mom. My dad says nothing because he makes me more upset.* (Adult with PWS 003) *– I watch television, I listen to music and I pray every day. Every morning I make the beds, and I set the table while my parents are cooking.* (Adult with PWS 012)
Outdoor activities	*– I refresh myself by being alone. I do yoga, I study Japanese, I sing in a choir. I have not given up my passions.* (Caregiver 074) *– As the years went by, I dedicated more and more time to my son. So, I gave up my passions to devote myself to him. I still cultivate a passion for sport, which I rarely practice. I love reading at night.* (Caregiver 019) *– I would like to swim, to go for walks by the sea and around the lake. I would like to have no responsibilities, schedules or limitations; I would like to be a little carefree.* (Caregiver 012)	*– I do athletics, especially playing football and swimming. I go for walks with others, alone or with my dog. I like going to see my favourite football team play at the stadium. I play videogames and watch movies. I like going to see my favourite animal, the dolphin, at the dolphinarium.* (Adult with PWS 011) *– I try to help people in trouble. I do a lot of activities such as swimming pool, occupational therapy and kinesiotherapy because I have had back surgery twice for severe scoliosis.* (Minor with PWS 006) *– I like going out, shopping, swimming, listening to music, drawing, cutting and pasting, making figurines and making collages.* (Adult with PWS 027)
Relationships	*– Some people have tried to understand the condition, but others have disappeared. The syndrome helped me to choose from among the people around me.* (Caregiver 075) *– My father and my mother were as sorry and as much in disbelief as I was, and they were unable to get over it. My sister and friends supported me. My husband shut himself off,*	*– My parents are the closest and most valuable people. They are strict but affectionate and loving. I am always looking for contact with them, even if I have made them angry. My brothers are also close to me. One of them is playful, and the other one is protective, a little severe. With them, I am calm and feel protected.* (Adult with PWS 038)

	and he is present and absent at the same time, except for practical things. (Caregiver 004) *– The grandparents have been fantastic, available and always ready to help us. Our friends did not understand our situation, and our roads soon split: your social life changes radically.* (Caregiver 066)	*– At school, I have tried to get closer to others, but they do not like to be with me and do not want me. My professors love me; however, with some of them, I feel that I am different and not good. As for my parents, my biological mother did not love me, and my father hated me; my new parents love me so much.* (Adult with PWS 014) *– I love my family. They do everything they can to make me happy.* (Minor with PWS 008)

Work and future perspectives

Sixty-two percent of family caregivers had to change their job after the birth of their child with PWS (Table 6): more than one third left their current work, 8% changed jobs to assist the child, 8% requested a part-time job, and 3% abandoned the perspective of a career. From a gender perspective, 63% of female (mothers) and 33% of male caregivers had to change their current job to adapt to the child's condition. Forty-six percent did not discuss PWS in the workplace or discussed PWS with only their closest colleagues; 36% reported comprehensive behaviours, and 18% declared a lower understanding of PWS management necessities than for other diseases, such as cancer. Based on the narratives, work was a positive personal resource. Fifty-six percent of adults with PWS declared that they were working in jobs mainly characterised by low complexity and repetitive operations and conducted in social cooperatives or centres, small companies with high corporate social responsibility, and family companies. Work is a source of pride and well-being and a distraction from food, but episodes of irritability and aggressive behaviours have been reported.

Table 6 – Prader–Willi syndrome and the work sphere: quotes from narratives

Adults with PWS	*– My colleagues are supportive; they are good listeners and give me good advice.* (Adult with PWS 008) *– I deal with publicity, I answer the phone, I receive the people who visit, and I do other tasks that the office managers assign me. If I am happy and motivated, I do not go looking for food. I look for food only when I am nervous. I have made so much progress, and so I do it less.* (Adult with PWS 015) *– I often have nervous attacks and respond badly. I offend with swear words those who make fun of how I am doing my job.* (Adult with PWS 006) *– I work in a nursing home 5 days a week. I wash laundry and iron and fold the guests' clothes. I work with a girl who takes care of me and teaches me many things. Every day I work with a different person, depending on the shift. With them, I feel good*

	because they treat me like a normal person, and they praise me every time I work with them. (Adult with PWS 013)
PWS caregivers	*– My colleagues firmly support me. They give me the ability to meet my personal needs, which allowed me not to leave my son and my wife alone. They were like a second family to me, and they all contributed as much as they could.* (Caregiver 054) *– I had to leave work. I was working in the factory 8 hours a day, and it was not possible to ask for a part-time position.* (Caregiver 053) *– The job was a safety valve, a place to have a normal life.* (Caregiver 023) *– When another daughter of mine had cancer, my colleagues were very involved, helpful and supportive. Everyone understands cancer. The disability is different, particularly the Prader–Willi syndrome, which is not very obvious.* (Caregiver 008)

Regarding future perspectives, PWS family caregivers hope to have long lives so that they can care for their sons and daughters as long as possible, and they were particularly concerned with what would happen to their children without familial support (Table 7). Caregivers also addressed social inclusion, such as social changes and openness, rather than clinical solutions to PWS. Adult participants with PWS demonstrated self-realisation through work (27%)—as underaged participants did—and the desire to have a family (46%), recover from PWS (10%), and generally live well (14%).

Table 7 – Living with Prader–Willi syndrome and future thoughts: quotes from narratives

Minors with PWS	*– When I grow up, I want to be a professor of endocrinology to study my syndrome.* (Minor with PWS 007) *– I want to be a vet because I love animals.* (Minor with PWS 014) *– I want to drive an ambulance and do many different jobs.* (Minor with PWS 009)
Adults with PWS	*– I want to start a family by adopting a child because I have so much love to give and to be with my boyfriend. I hope to work in the same place where I am. I do not know how to live without my parents; they help us a lot.* (Adult with PWS 013) *– I want to go live with the person I love the most in the world and to have children. Above all, I want to become a famous singer, and personally meet all my favourite singers. I want to get married and live in a beautiful house with a pool.* (Adult with PWS 009) *– I hope that medicine will be able to find a drug to that can increase my satiety and limit my nervous hunger, which is unfortunately increased by my syndrome. Some moments are hard to overcome. I hope to live as well as I can with the consequences of the syndrome, and I hope that doctors are able to find treatments to alleviate the challenges. I hope that other people will also be able to live better lives.* (Adult with PWS 015)
PWS caregivers	*– I am sure that his future will be full of satisfaction. We are working to ensure a positive future for him and to find him a job that makes him happy and confident in his abilities.* (Caregiver 075) *– I believe the future will be challenging; I hope that I am wrong, but I realised that most people care most about their own business. Indeed, it is painful and challenging to care for or remain close to people with this kind of pathology. Still, perhaps if people did their part and committed themselves more to social causes, the future could be different. We should learn from an early age that we need to help other people and stand by those in need.* (Caregiver 054)

> *– As long as we, her family, are there, I think that her future is protected. What comes after is frightening, but I know I have to do something for her ... I owe her that!* (Caregiver 031)

DISCUSSION

The PRAXIS project represents the first effort to investigate the PWS illness experience in Italy through NM by simultaneously considering the perspectives of underaged and adult patients with PWS and their caregivers. The project first aimed to understand their daily life, real needs and personal resources. Fifty-five participants, namely, children, adolescents and adults with PWS, reported joy and pride in sharing their stories, also suggesting that using evocative and open words[36] in structuring illness plots can be crucial to helping people to express themselves. Moreover, the collection of 138 caregiving stories suggested a strong dedication to the survey and the need for caregivers to be listened to: They described writing as liberating, demonstrating its potential (a) to have a therapeutic effect[27, 33] and (b) to be a safe space from the attitude of passing,[41] namely, handling information considered discrediting or critical for the self to avoid social stigma.

Talking about PWS emerged as a 'taboo'. In illness-centred and sickness-centred narratives [28], caregivers encountered significant difficulties in socialising the challenges PWS imposes in daily life, as well as the pain of having a child "different" from social imagery: We identify this as a social pain that also concerns caregivers when performing familiar criticalities. Furthermore, caregivers considered the project a chance to invite society to integrate people with PWS and to denounce the stigma[41] that surrounds them.

If the literature demonstrates cognitive impairment in people with PWS,[6] we would like to enrich the evidence by suggesting the consideration of the multiple intelligences[42] these people demonstrate in their everyday experience. In line with Gardner's[42] reflection, revealing alternatives to the standard forms of intelligence (the logical-mathematical and linguistic ones),

the narratives demonstrated the constant use of visuospatial, musical, interpersonal, existential and introspective talents, resources and capabilities. In this regard, patients with PWS have been reported to show above-average performance in several tasks implying visuospatial skills,[43] which in the general population are linked with higher math abilities.[44] In particular, the importance of multiple intelligences emerged in food- control strategies and activities, consequently suggesting that considering them may positively influence the overall illness experience.

Considering the second purpose of the project, specific elements emerged from the analysis of therapeutic paths. PWS diagnosis mostly occurred up to the child's second year of life, but in some cases, a significant delay remained, particularly for those people living away from specialised centres. Because timing is essential in PWS treatment,[9] training for neonatologists, geneticists and general paediatricians on PWS might improve early diagnosis. Moreover, the other professionals involved also must develop or strengthen specific PWS competencies to appropriately address this condition.

The narratives demonstrated some peculiar clinical PWS characteristics, such as irritability, aggressive and obsessive-compulsive behaviour [10, 11] and food-seeking[13] behaviours. The last resulted in the most challenges for the people with PWS and their caregivers: Food management strategies,[45] and indoor and outdoor activities[46] and school or work schedules,[47, 48] can help people with PWS and their caregivers improve their relationships with food and family members, enhancing overall daily life.

Diet management and strategies, early GH therapy, clinical and psychiatric treatments, activities in specialised or social centres, and the different professional roles involved demonstrated that a mutiprofessional approach that integrates the factors of hospital and territory is fundamental to ensure adequate treatment of PWS [17, 47, 49] and to mitigate the burden of caregiving reported

in the narratives and the literature.[14-16] In particular, two related topics emerged: (a) mostly women (mothers) changed or retired from work to become a caregiver; (b) family caregivers stated their concerns regarding what will happen to their sons and daughters if no family members are available —a topic already addressed by Italian Law 112/2016, on the social inclusion and autonomy of people with disabilities. These considerations also suggest, on the one hand, that social centres and services are crucial but need to be implemented in areas that have insufficient support for people with PWS and their caregivers, and on the other hand, a focus on work policies to create autonomy and social inclusion. Overcoming economic, legal, and social barriers and improving the current service provision still represent a challenge; patient organisations and scientific societies may have a crucial role in addressing these issues. Furthermore, although a national plan for rare diseases[50] has been developed in Italy since 2013, its application in daily practice remains demanding. One possible intervention strategy to reduce medical barriers requires universities and scientific societies to develop specific educational programmes; in particular, creating a PWS national register may help to interface with similar international tools.[51]

The acknowledgement of the importance of multiple intelligences[42] in everyday experiences may also improve the daily and relational life of people with PWS or their caregivers, together with ameliorating the social stigma of PWS and enhancing social inclusion. Multiple intelligences might also become a tool in clinical practice to better evaluate people with PWS; moreover, an evolving model for PWS care should include modern technologies, e.g. video visits, remote monitoring and electronic health records.[52]

The participants in the project did not equally represent the different geographical areas in Italy because of the local distribution of expert centres in the management of care for PWS, and this could be a selection bias. Furthermore, the results are specific because of (a) the voluntary nature

of the project and (b) the critical difference among Italian regional healthcare systems; therefore, further analysis is required. For people with PWS, the inclusion criterion of being able to write represented another bias.

In conclusion, this NM project provides new insights into the individual and social experiences related to PWS and provides elements for improving multidisciplinary and multiprofessional perspectives on this condition: The social, relational, and emotional aspects of PWS crucially influence the illness experience and narratives that can foster the relationship between PWS professionals, patients, families and the community.

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Competing interests

Authors have nothing to disclose.

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Author contributions

Conceptualisation: Ragusa, Crinò, Grugni, Reale, Marini. Analysis: Reale, Fiorencis. Investigation: Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Methodology: Reale, Marini. Project administration: Reale. Ragusa, Crinò, Grugni, Reale, Marini. Report visualisation:

Ragusa, Crinò, Grugni, Reale, Fiorencis, Licenziati, Faienza, Wasniewska, Delvecchio, Franzese, Rutigliano, Fusilli, Corica, Campana, Greco, Chiarito, Sacco, Toscano, Marini. Writing: Fiorencis, Ragusa, Crinò, Grugni, Reale, Marini. Editing: Fiorencis.

Data sharing

All data relevant to the project are included in the present manuscript. Original narratives are available into Italian upon request at the email areasanita@istud.it.

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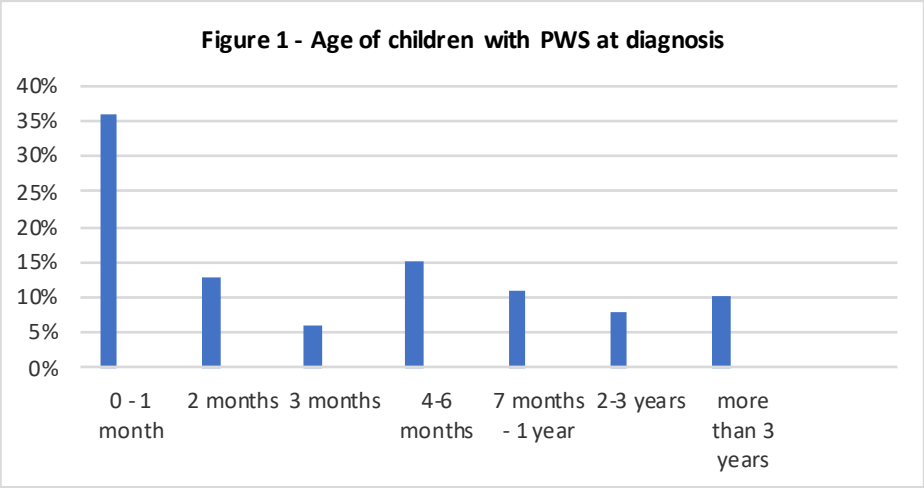
Figure legend

Figure 1 – Age of children with PWS at diagnosis



Supplement 1

Medical centres for paediatric and adult patients of the Italian Network for Rare Disease

OASI Maria SS., Research Institute, Troina (Enna), Italy

Bambino Gesù Paediatric Hospital, Research Institute, Palidoro (Rome), Italy

Istituto Auxologico Italiano, Research Institute, Piancavallo (Oggebbio, Verbania), Italy

Obesity and Endocrine Disease Unit, Department of Neuroscience, Santobono-Pausilipon Children's Hospital, Naples, Italy

Department of Biomedical Sciences and Human Oncology, University of Bari "A. Moro", Bari, Italy

Department of Human Pathology of Adulthood and Childhood "G.Barresi", University of Messina, Italy

Metabolic Diseases, Clinical Genetics and Diabetology Unit, Giovanni XXIII Children's Hospital, Bari, Italy

Department of Translational Sciences, University Federico II, Naples, Italy

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Neonatology Unit, Spirito Santo Hospital, Pescara, Italy

Supplement 2

Illness plot addressed to underaged people with PWS

Hi! Can you tell us about yourself? […] May you introduce yourself and tell us about your life [...]. How do you feel with your friends […], your schoolmates […], and teachers? [...] You know that eating too much is not good for you: what do you do for not eating too much? [...] How do you feel with your family? […] Can you tell us about your visits to the doctors? […] What is your favorite game? […] What will you do when you grow up? [...]

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to adult people with PWS

I introduce myself [...]. I feel [...]; at home [...]; at work [...]. The people next to me [...]. The food [...]. The treatments [...]. What I like to do [...]. For me, I think of a future [...].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Illness plot addressed to caregivers of people with PWS

When I felt the first signs that something was going wrong […] I feel […]. They told us that it was […]. The hospitals I visited, the professionals I met […]. That period at home […], at work […]. The people next to me [...]. People around the person with PWS […]. Food […], and treatments […]. Today the person with PWS […], and treatments […]; I feel […]. At home, people with PWS […]. Outside domestic context, the person with PWS […]. What I like to do […]; what the person with PWS likes, beyond food […]. My life at home is […]; the food […]. I look at the person with PWS and I think for her/him a future […], and for me I imagine a future […].

Thank you for the time and energy you dedicated. One last question:

How did you feel in narrating your experience? [...]

Reporting checklist for qualitative study.

Based on the SRQR guidelines.

Instructions to authors

Complete this checklist by entering the page numbers from your manuscript where readers will find each of the items listed below.

Your article may not currently address all the items on the checklist. Please modify your text to include the missing information. If you are certain that an item does not apply, please write "n/a" and provide a short explanation.

Upload your completed checklist as an extra file when you submit to a journal.

In your methods section, say that you used the SRQRreporting guidelines, and cite them as:

O'Brien BC, Harris IB, Beckman TJ, Reed DA, Cook DA. Standards for reporting qualitative research: a synthesis of recommendations. Acad Med. 2014;89(9):1245-1251.

		Reporting Item	Page Number
Title			
	#1	Concise description of the nature and topic of the study identifying the study as qualitative or indicating the approach (e.g. ethnography, grounded theory) or data collection methods (e.g. interview, focus group) is recommended	1
Abstract			
	#2	Summary of the key elements of the study using the abstract format of the intended publication; typically includes background, purpose, methods, results and conclusions	2
Introduction			

Problem formulation	#3	Description and signifcance of the problem / phenomenon studied: review of relevant theory and empirical work; problem statement	3-4
Purpose or research question	#4	Purpose of the study and specific objectives or questions	5
Methods			
Qualitative approach and research paradigm	#5	Qualitative approach (e.g. ethnography, grounded theory, case study, phenomenolgy, narrative research) and guiding theory if appropriate; identifying the research paradigm (e.g. postpositivist, constructivist / interpretivist) is also recommended; rationale. The rationale should briefly discuss the justification for choosing that theory, approach, method or technique rather than other options available; the assumptions and limitations implicit in those choices and how those choices influence study conclusions and transferability. As appropriate the rationale for several items might be discussed together.	4-6
Researcher characteristics and reflexivity	#6	Researchers' characteristics that may influence the research, including personal attributes, qualifications / experience, relationship with participants, assumptions and / or presuppositions; potential or actual interaction between researchers' characteristics and the research questions, approach, methods, results and / or transferability	7
Context	#7	Setting / site and salient contextual factors; rationale	5-6
Sampling strategy	#8	How and why research participants, documents, or events were selected; criteria for deciding when no further sampling was necessary (e.g. sampling saturation); rationale	6
Ethical issues pertaining to human subjects	#9	Documentation of approval by an appropriate ethics review board and participant consent, or explanation	7-8

		for lack thereof; other confidentiality and data security issues	
Data collection methods	#10	Types of data collected; details of data collection procedures including (as appropriate) start and stop dates of data collection and analysis, iterative process, triangulation of sources / methods, and modification of procedures in response to evolving study findings; rationale	7
Data collection instruments and technologies	#11	Description of instruments (e.g. interview guides, questionnaires) and devices (e.g. audio recorders) used for data collection; if / how the instruments(s) changed over the course of the study	7
Units of study	#12	Number and relevant characteristics of participants, documents, or events included in the study; level of participation (could be reported in results)	9
Data processing	#13	Methods for processing data prior to and during analysis, including transcription, data entry, data management and security, verification of data integrity, data coding, and anonymisation / deidentification of excerpts	8
Data analysis	#14	Process by which inferences, themes, etc. were identified and developed, including the researchers involved in data analysis; usually references a specific paradigm or approach; rationale	8
Techniques to enhance trustworthiness	#15	Techniques to enhance trustworthiness and credibility of data analysis (e.g. member checking, audit trail, triangulation); rationale	8
Results/findings			
Syntheses and interpretation	#16	Main findings (e.g. interpretations, inferences, and themes); might include development of a theory or model, or integration with prior research or theory	9-10
Links to empirical data	#17	Evidence (e.g. quotes, field notes, text excerpts, photographs) to substantiate analytic findings	10-16

Discussion			
Intergration with prior work, implications, transferability and contribution(s) to the field	#18	Short summary of main findings; explanation of how findings and conclusions connect to, support, elaborate on, or challenge conclusions of earlier scholarship; discussion of scope of application / generalizability; identification of unique contributions(s) to scholarship in a discipline or field	16-19
Limitations	#19	Trustworthiness and limitations of findings	19
Other			
Conflicts of interest	#20	Potential sources of influence of perceived influence on study conduct and conclusions; how these were managed	20
Funding	#21	Sources of funding and other support; role of funders in data collection, interpretation and reporting	20

None The SRQR checklist is distributed with permission of Wolters Kluwer © 2014 by the Association of American Medical Colleges. This checklist can be completed online using https://www.goodreports.org/, a tool made by the EQUATOR Network in collaboration with Penelope.ai