REVIEW

Patient journey experiences may contribute to improve healthcare for patients with rare endocrine diseases

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Abstract

Patient journeys are instruments developed by EURORDIS, The Voice of Rare Disease Patients in Europe, to collect patients' experiences; they may identify gaps and areas deserving improvement, as well as elements positively considered by affected persons. As with other patient-reported experiences, they can complete the clinical evaluation and management of a specific disease, improving the often long diagnostic delay, therapy, patient education and access to knowledgeable multidisciplinary teams. This review discusses the utility of such patient-reported experience measures and summarises the experiences of patients with acromegaly, Addison's disease and congenital adrenal hyperplasia from different European countries. Despite rare endocrine diseases being varied and presenting differently, feelings of not having been taken seriously by health professionals, family and friends was a common patient complaint. Empathy and a positive patient-centred environment tend to improve clinical practice by creating a trustworthy and understanding atmosphere, where individual patient needs are considered. Offering access to adequate patient information on their disease, treatments and outcome helps to adapt to living with a chronic disease and what to expect in the future, contemplating the impact of a disease on patients' everyday life, not only clinical outcome but also social, financial, educational, family and leisure issues is desirable; this facilitates more realistic expectancies for patients and can even lead to a reduction in health costs. Patient empowerment with patient-centred approaches to these complex or chronic diseases should be contemplated more and more, not only for the benefit of those affected but also for the entire health system.

Key Words

- health perception
- patient journey
- Addison's disease
- acromegaly
- congenital adrenal hyperplasia

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Introduction

Knowledge on patient experience related to the healthcare systems is currently considered essential to pursue optimal quality of care (1). The concepts of patient-reported outcome measures and patient-reported experience measures (PREMs) are nowadays considered more and more important in healthcare evaluation; in fact, improved PREMs are shown to be associated with the enhancement in the quality of healthcare, better financial outcome of health institutions and is even a predictor of survival (1, 2, 3, 4, 5). Patients especially associate a better outcome to the level of communication with and trust in their doctors and nurses, as well as sufficient information and understanding of outcome (4, 7). 'Patient journeys' (PJ) are instruments developed to collect information on patients' experiences in many areas of medicine, including rare diseases (8). They are simple ways to make visible the needs of a community of patients that can complete the view of the clinician on a specific disease if they wish. It is a way to share and discuss information between patients and clinicians, not only medical or on the natural history of the disease but also emotional, psychological, social and other issues that determine everyday needs. It enables a discussion to identify unmet needs and approach problems in order to try to solve them. These problems or gaps may be common for a group of patients with a certain diagnosis, as well as others specific to individual syndromes.

PJ were developed by EURORDIS, in collaboration with the ERN GENTURIS (the European Reference Network for patients with one of the rare genetic tumour risk syndromes; www.genturis.eu), after a mapping exercise of the needs of different rare inherited syndromes (8). It allows to connect professional expert guidelines (which include defined medical interventions, screening, treatment, etc.) with patient needs, not only medical but also psychological.

One of the main aims of the ERNs since their implementation in 2017 is to tackle the current hurdles of complex or rare diseases and to connect collective experience and expertise across Europe, facilitating the knowledge to travel instead of the patient. Collecting data on PJ from different countries with distinct and varying health system organizations, as well as from different diseases, may contribute to provide PREM information which is currently mostly lacking.

For patients, elements like functional status after a health intervention or treatment (like the presence or absence of pain, extreme fatigability, capacity to concentrate, etc.) are usually more important for their everyday life than more technical parameters usually used to measure health outcomes (i.e. mortality, complication rates) (9, 10). Additionally, next to elements of quality of healthcare, like safety and clinical effectiveness, elements like compassion, dignity, respect, unmet needs, gaps, and issues related to social, working capacity and other issues are important for patients, and not always contemplated by health administrators. Contemplating these elements with empathy from the healthcare professionals and providing information as a PJ document, be it a visual diagram or a brief text, can reduce suffering and can be mutually beneficial (1). Naturally, the exact content, scope and extension may vary, depending on the different issues stated earlier.

For patients with rare diseases, a long diagnostic delay and visits to many healthcare professionals before a correct diagnosis is reached may determine suffering and anxiety for years; therefore, when a diagnosis is finally reached, their degree of satisfaction is high, especially when additional support and information on their disease and consequences are available (7, 11). Neonatal screening may shorten the time to diagnosis for some diseases such as congenital adrenal hyperplasia and thereby prevent sequelae or neonatal death, in addition to the psychological aspects of avoiding diagnostic delay.

Concerns about data privacy are often put forward by some health professionals and legal bodies when asking patients for their opinions. However, PREMs are opinions voluntarily offered by affected patients and a good way to gain information on what it is like to be living with a chronic or rare disease; potentially, they can help the healthcare professionals to improve patient outcome and experience, beyond the more technical issues, creating a positive patient-centred, empathic environment. Furthermore, a recent systematic review of patient privacy perspectives related to health information exchange has evidenced that perceived quality of care was associated with few privacy concerns; in other words, positive patient perceptions on healthcare and exchange of health information reduce privacy concerns and its effects (12).

Other features which are important to relieve anxiety and suffering are to clarify the confusion and uncertainty of patients and their families. Waiting for a consultation can be very stressful, as well as having received conflicting information from different handling clinicians. Making relevant information easily available for new patients and good healthcare professional–patient communication is necessary to improve perception of the situation, as well as reminding the patient that coping is essential, despite the presence of the disease and its possible consequences,

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and that some degree of improvement is nearly always possible (11, 13, 14).

The lead of Work Package 4 (responsible for Quality of Care and Patient View) within the European Reference Network devoted to rare endocrine conditions (EndoERN) adapted the original template developed by EURORDIS to these endocrine conditions (Supplementary Table 1, see section on supplementary materials given at the end of this article).

Patient journeys within RareEndoERN

With this review, we wish to highlight the utility of PJ to gain further insight on the impact of a rare endocrine disease on an affected patient or their families. While medical textbooks and guidelines collect relevant aspects related to the diagnosis and management of a specific disease, they do not always include the patients' point of view, which can be reflected in different PJ formats (e.g. a visual diagram, a list of problems encountered and how to approach them, confront positive and/or negative experiences, or describe unsolved gaps or practical hurdles in everyday life).

Additionally, we performed a practical exercise of identifying around half a dozen patients with either acromegaly, Addison's disease or congenital adrenal hyperplasia and asked them to write down their experiences in a table template, initially created by EURORDIS, and reviewed and adapted by the authors for endocrine patients.

Patients from the countries of the WP4 leads were initially approached by identifying a representative patient for three diseases related to the main thematic group 1 (MTG1: adrenal) and MT6 (pituitary) of EndoERN, affecting mainly children (congenital adrenal hyperplasia) or adults (Addison's disease and acromegaly). After explaining the aim, they volunteered to collaborate and were sent the template table to fill and return to one of the four WP4 leads. They were encouraged to discuss their opinions with other patients or associations they are related to, with the idea of getting a realistic and full picture of what they have experienced. Before completing any data, patients confirmed they had read and understood the attached 'Information on Collecting Data for Participation in Patient Journey' (Supplementary Table 1) and consented to the processing of the reported data as described therein. From the original English version, translations have been performed by the authors into Danish, French, German, Italian, Spanish and Swedish (Supplementary Table 2).

Patient journey in acromegaly

Data of six patients from four countries were obtained (Tables 1 and 2). Diagnostic delay oscillated from 3 months to 15 years after the appearance of symptoms. However, most symptoms were only recognised retrospectively, once the diagnosis had been reached. Typical clinical presentation and symptoms, both physical and psychological, had for some patients been experienced for years but were unrecognised by different healthcare professionals (11). Less recognised features like a very large placenta which was mentioned but not interpreted as diagnostic, or a laryngeal mucosal hypertrophy determining upper airway narrowing, recognised by an experienced ENT surgeon who had diagnosed another patient in the past based on the same finding, after consulting for a dysphonia (15), deserve highlighting. Another patient-referred feature was 'a change in smile', obviously hardly perceivable by anyone who is not the patient or in their close circle.

Patients expressed frustration in not having been listened to when complaining that 'something was wrong', and were not taken seriously, or had to insist repeatedly to be referred to a reference centre, fighting many administrative battles. A desire to understand what was going on, with a plausible explanation and to feel at peace was verbalised; access to quality information and care was highlighted, as well as an empathetic attitude, ideally by multidisciplinary teams who could offer global care, contemplating the disease itself and all accompanying morbidity. Access to experienced neurosurgeons, without a long waiting list, was stated as important. However, this was only possible in private practice in some cases and not accessible to all. Clarifications of 'scary' words like having a tumour in your head or requiring radiotherapy (associated to cancer) were stated as essential for the patients.

Table 1 Available translations and countries in which the patient journey template for different diseases was completed.

	Denmark	France	Germany	Italy	Lithuania	Spain	Sweden
Diseases							
Acromegaly	x	x			x (in English)	x	
Addison's disease	x			x		x	x
Congenital adrenal hyperplasia			x	x			x

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Table 2 Summary of patient journey data in acromegaly

Stage of journey	**Clinical presentation/symptoms**	**Patient/family needs**	**Ideal outcome/support** (how to address the needs)
Pre-diagnosis: Diagnostic delay of 0.25–15 years before correct diagnosis First symptom or at diagnosis	Typical physical: headache, carpal tunnel sd, sweating, menstrual irregularity, feeling tired with less strength, growth of hands, feet & tongue (determining lisping), joint pains, facial changes perceived suddenly by third parties, appearance of more body hair, snoring or sleep apnea, weight gain, skin thickening, jaw growth with toothache, high blood pressure Feeling 'something is wrong' Psychological: insomnia, lack of concentration, less memory, unreasonable nervousness, depression, fits of rage, irritability Less frequently described: 'unexplained' very large placenta. Laryngeal ventricular hernia, change in smile	Two different scenarios: - No suspicion of disease, problems thought to be related to stress, ageing, weight gain or lifestyle changes - Years of repeated consultations with no recognition of the symptoms as apart of the disease, determining stress and impaired QoL Desire to understand what is going on and feel at peace Fear of reaction of relatives when disclosing the diagnosis High-quality information, care & empathy by specialised multidisciplinary reference centres to offer global care. Accessibility to experienced neurosurgeon for all (not only to expensive private specialists) Honesty by HCP if not familiar with the disease, to refer to reference centre Information on radiotherapy, associated to 'cancer' and scary Earlier recognition by sleep centres, neurologists, ENT specialists, dentists, gynaecologists, orthopaedic surgeons, jewellers, shoe retailers, etc. Understanding that it is a chronic life-changing disease and learn to live with it, in a positive way	Listening to the patient by all involved in their healthcare (primary care, specialists consulted, nurses) who complain they are not taken seriously despite knowing 'something is wrong'. Increase awareness of the disease at all health levels to reach an earlier diagnosis and prevent irreversible changes Empathy by primary care and prevent delays in referring patients or organising diagnostic tests Access to multidisciplinary expert teams not only for diagnosis but also for chronic follow-up Shorten waiting list for surgery, which is lived as very stressful. Empowering patient associations, since sharing experiences can be very therapeutic, leading to better information, acceptation and peace Access to psychiatrist or psychologist who understands the psychological aspects of the disease Utility of questionnaires on QoL or different aspects of the disease which help the patient realise that they are validated and not crazy

Most referred symptoms and quality of life improvements after surgery or medical therapy, but some problems persisted and impaired their quality of life. Long delays to receive treatment were mentioned as generators of great stress and anxiety, while a sensation of great relief was experienced after surgery.

As far as patient or family needs, two different scenarios were reported. While for some there was no suspicion of any abnormality and perceived problems were considered to be related to stress, ageing, weight gain or unhealthy lifestyles, for the majority, stress and impaired quality of life were perceived after years of multiple and varied consultations due to different health problems, without recognition of the disease. These included sleep specialists, neurologists, ENT surgeons, dentists, gynaecologists or orthopaedic surgeons, and also jewellers or shoe retailers whom it is worth to be made aware of the extreme rarity of significant increase in hand or foot size, if not due to acromegaly.

The positive role of patient support associations to share experiences and listen to other patients who had lived similar situations was considered very therapeutic by many, leading to relevant information, acceptation, coping and a sensation of peace. Given the psychological implications of acromegaly, many valued the possibility of access to a psychiatrist or psychologist to complete their global management. Becoming aware by questionnaires that many complaints were related to the disease was also stated as helpful since they felt validated that what they had experienced was real and not their imagination or becoming a hypochondriac.

Finally, understanding that acromegaly is a chronic disease, with long-term consequences and requiring in some cases monthly injections and frequent contact with the healthcare system, is essential. Positively adapting to the life changes required is important and will help come to terms with the situation, while maintaining a satisfactory daily quality of life.

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Patient journey in Addison's disease

Data of six patients from four countries were obtained (Tables 1 and 3). Patients referred having suffered from symptoms related to the disease from 6 months to 10 years before diagnosis, mostly from 2 to 4 years. These symptoms were those classically described for adrenal insufficiency but not recognised as such by the healthcare professionals consulted, namely nausea, vomiting, fainting, extreme tiredness, loss of weight, anorexia, hypotension, shortness of breath on minor exertions like climbing stairs, pigmentation of skin, hand line and lips, menstrual irregularity in women, often leading to anxiety and claustrophobia. Additionally, some patients referred to salty food craving and worsening of all symptoms after minor health problem like a cold, an infection or a surgical procedure. Once recognised, however, all stated progressive physical and psychological recovery as soon as glucocorticoid and mineralocorticoid substitution therapy was initiated. Individualisation of the total daily dose and distribution throughout the day was not always immediate, highlighting the need to discuss each patient's usual wake/sleep cycle and food and activity timetables to

Table 3 Summary of patient journey data in Addison's disease

Stage of journey	**Timeline**	**Clinical presentation/symptoms**	**Patient/family needs** (collective view)	**Ideal outcome/support** (how to address the needs)
Pre-diagnosis very symptomatic. After diagnoses and start of treatment, great improvement although finding the exact dose required and its daily distribution may take some time	2 years 10 years 6 months 3–4 years 1–2 years	Nausea & vomiting, fainting, extreme tiredness, loss of weight, anorexia, shortness of breath on minor exertions like climbing stairs, skin pigmentation, brown hand lines, 'blue' lips, anxiety and claustrophobia, hypotension, menstrual disappearance Salty food craving, Exacerbation after minor health problem like a cold, any infection or surgical procedure Progressive physical and psychological recovery after diagnosis and starting glucocorticoid and mineralocorticoid substitution therapy. May take some time to find the correct daily dose and distribution	No understanding by family would appreciate more listening to the patients problems and concerns Disease not recognised by primary care Desire to recover lost energy and improve the shortness of breath It was good to get a diagnosis and not feel like a hypochondriac who was always in pain and tired Necessity of explanations on what it means to live with a chronic, life-long disease Education on physical and psychological situations that require extra stress-dose increase in glucocorticoid	Finding medical staff who listen to the patient and are knowledgeable on the disease. Relief after clear diagnosis and required therapy Education and access to knowledgeable specialists Information and explanations for family who have difficulties in grasping what the disease entails, how to live with it and what to expect Awareness of the need to adapt the daily treatment to personal timetable and activity, understanding the underlying physiology Awareness and information of other endocrine deficiencies which may accompany Addison's disease: hypothyroidism, vitamin B12 deficiency, premature menopause. Have a stable follow-up team, with the same doctor or nurse, and not constant changes that create anxiety during follow-up Training on crises management requiring dose increase and injections in case of infection, exertion or any emergency situation Better physician training on emergency care and crises management with immediate administration of parenteral glucocorticoids Access to constant medical support if required - Facilitate access to hydrocortisone at all levels (not only in the hospital pharmacy, as occurs in Italy)

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optimise therapy and well-being. Being aware of sick day rules, when doses should be immediately increased, as well as education on parenteral administration of glucocorticoids if the oral route is not possible, was also considered essential (16).

As far as patient and family needs, they were in part common to those expressed by patients with acromegaly. Despite both being rare endocrine diseases, they have little in common as far as symptoms are concerned, but both stress the importance of access to reliable education for the patients and family on the nature of the disease, what it entails as a chronic life-long procedure, as well as the initial relief of knowing that finally their complaints were explained and validated, after a long time of not being understood by their family or those healthcare professionals consulted. For the patient, recovering their energy and feeling less pain once glucocorticoids were given was a tremendous relief.

Additionally, being aware that Addison's disease may often be part of an autoimmune polyendocrinopathy with additional hypothyroidism, premature menopause and/or vitamin B12 deficiency requiring monthly injections was also considered important to accept the situation and find alternatives.

Despite the medical textbook knowledge of the need for immediate parenteral glucocorticoid administration when patients require an emergency consultation, this is often not the case; unjustified delays are a common complaint in many patients and relatives, despite informing of this immediate need, especially when non-endocrine specialists are involved. This is clearly a gap which deserves improvement at all levels.

A stable follow-up with the same personnel was stated as highly reassuring and desirable, if possible.

Finally, in Italy, there was the extra complication of only having access to parenteral hydrocortisone at hospital pharmacies, and patients plead to make it easier and more accessible.

Patient journey in congenital adrenal hyperplasia

Data of seven patients from three countries were obtained (Tables 1 and 4); in one of the Italian reports, two affected girls from the same family were reported. While the first daughter was born with ambiguous genitalia and salt wasting and immediately admitted to the neonatal ICU unit where she was treated and promptly diagnosed, the second daughter was diagnosed after a chorionic villous biopsy at 11 weeks of pregnancy. Her mother was treated with glucocorticoids throughout pregnancy so that at birth genitalia were normal and treatment with glucocorticoids was initiated. The first daughter, however, required surgery for external genitalia correction at the age of 4 months.

Another case was also diagnosed at birth while in another two it took 2–3 weeks for the correct diagnosis to be made. This created great anxiety in the parents, who complained often of not being taken seriously or having the personal and administrative problem of not being sure of the gender assignment of the newborn. All insisted on the need for speedy confirmation of diagnosis at birth or immediately after. One of the mothers complained of having been left alone in the delivery room without any explanation which created great anxiety, while the newborn and father were taken to the neonatal ICU facilities.

A boy identified via neonatal screening was reported by the parents. Symptoms of irritability and insufficient weight gain did not give the diagnosis before the screening result at 8 days. He was admitted to the hospital and started on treatment the same day. The parents describe difficulties taking in the information and understanding the disease and its consequences and would have needed more support in how to give the medication and the salt to their baby.

A further male patient who did not have salt-wasting was diagnosed at the age of 2½ years, due to androgen excess, determining excessive growth, advanced bone age and pubic hair. The risks of both under- and over-replacement with gluco- and mineralocorticoids were mentioned, which could be minimized if all patients were cared for by reference multidisciplinary groups, where a clear treatment plan and care pathway was established and agreed upon by the healthcare providers and the families. Again, the desirability and positive influence of patient or family support groups was highlighted, as well as their inclusion in national disease registries, to make them easier to identify.

Similarly to the patients with Addison's disease, clear instructions as what to do when otherwise sick were considered essential, not only for the families but also for all health professionals involved (primary care, nurses, surgeons or any other healthcare providers, especially in the emergency room) (16). And easy access to parenteral hydrocortisone in all pharmacies (not only in the hospital) was a desire expressed in Italy. An on-call phone number in case of emergency, as well as to be able to ask questions and for less experienced physicians to get guidance on how to treat, was the kind of support that was requested.

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Table 4 Data from patient journey in congenital adrenal hyperplasia

Stage of journey	**Timeline**	**Clinical presentation/symptoms**	**Patient/family needs** (collective view)	**Ideal outcome/support** (how to address the needs)
Diagnosis	* 11 weeks of pregnancy 2 at birth 14 days postnatal 20 days postnatal **2.5 years of age	Extreme weakness, polyuria, vomiting, dehydration, acute salt loss, ambiguous genitalia (girls) *2nd daughter of affected family; mother treated throughout pregnancy and girl was born with normal genitalia **Non-salt wasting CAH diagnosed due to excessive growth, pubic hair, advanced bone age	More attentive clinical staff/ paediatrician to immediately recognise CAH, especially in salt-wasting cases. Being taken seriously, not as anxious patients Possibility of preventing ambiguous genitalia in a subsequent pregnancy by *in utero* genetic diagnosis and treatment of mother Speedy confirmation of diagnosis at or immediately after birth Awareness of all hospital staff of the need of hydrocortisone therapy at specific times throughout the day. Information on the physiology of normal cortisol secretion to understand the importance of adequate hydrocortisone dose with adequate circadian distribution (not too late at night which favours night-time waking). Risks of both under- and over-replacement treatment Information of the disease, what it entails as a chronic, life-long disease, which requires daily treatment, with a clear treatment plan and care pathway, both for parents and affected child Access to a knowledgeable specialist, rather than being handled in a trial-and-error fashion Patient/family support groups	-Access to empathetic and knowledgeable HCP who immediately recognise the diagnosis and begin therapy with hydrocortisone. -Rapid genetic diagnosis, especially if ambiguous genitalia, to register the child with a correct gender (required in 10 days in countries like Italy) - Sufficient information and support to patients to understand the disease and handle daily treatment and changes required if child is otherwise sick (sick day rules for medication). - Access to support groups and contact details of expert reference centres to optimise treatment - Adequate support and information in the delivery room for mother, father and child who may be rapidly transferred for further testing or admission to ICU - Facilitate access to hydrocortisone at all levels (not only in the hospital pharmacy, as occurs in Italy)
	Screening diagnosis 8 days of age Boy with SW form	Irritable, difficult to comfort, but no electrolyte disturbance. Insufficient weight gain not identified before the screening result	Important to receive clear and repeated information from doctor and the team. Difficult to take in all information and learn how to deal with the situation, both practically and psychologically. Support by nurses in how to give medication and especially the salt.	Repeated information and easy access to care. Good with a phone number to call in case of emergency that can give support also to other physicians in hospitals elsewhere. Same nurses and doctor over time, as far as possible, is important for feeling of security. Support in how to give the medication and especially the salt to a baby. Psychological support.

Conclusions

This exercise of highlighting the utility of and collecting patient-reported experiences as PJ in three different rare endocrine conditions has identified gaps where improvement in diagnosis, therapy, patient education and access to knowledgeable multidisciplinary teams are mentioned. Despite rare endocrine diseases being very varied and presenting differently (from mild to severe), the feelings of not having been taken seriously both by healthcare professionals and the patients' close circles of family and friends were rather common. When relevant, identification through screening shortens the journey to diagnosis and prevents some of the initial

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severe symptoms and possible sequelae (17). However, the importance of information and the needs in the context of long-term follow-up are also identified as very important. No information on patients' experiences regarding transition from childhood to adult care (a vulnerable period when being aware of patients' experiences and needs are of great importance) were included.

A further value of the PJ tool is that it can lead to create discussion and partnership between patients and clinicians for these rare diseases. Any derived findings of issues reported may then be used in EndoERN to propose new activities (i.e. in guideline development) or in new strategies, development of patient information and communication material.

Empathy and a positive patient-centred environment deserve pursuing since they can improve clinical practice, by creating a trustworthy and understanding environment; listening to the individual patients' needs is essential to attain positive patient experiences. This can be achieved by offering access to adequate patient information on the disease, treatments and outcome so that the patient knows what to expect in the future and adapt to living with a chronic disease. Further research devoted to analyse the impact of a disease on patients' everyday life, contemplating social, financial, educational, family and leisure issues, is desirable. In this way, those affected can contemplate realistic expectancies and correlate these issues with a reduction in health costs, related to both physical complaints and psychological ones like stress, depression and anxiety (18). Patient empowerment with patient-centred approaches to these complex or chronic diseases should be contemplated more and more, not only for the benefit of those affected, but also for the entire health system, as evidenced in a recent systematic review (19). Although the existing evidence was small to moderate, patient-mediated interventions, like patient-reported health information and patient education, were shown to improve professional practice by increasing healthcare professionals' adherence to recommended clinical practice.

Supplementary materials

This is linked to the online version of the paper at https://doi.org/10.1530/EC-22-0385.

Declaration of interest

The authors declare that there is no conflict of interest that could be perceived as prejudicing the impartiality of this review.

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Author contribution statement

All authors contributed and agreed to the final PJ template. SMW wrote the initial version of the manuscript which was edited and approved by all authors.

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References

1 Lee TH. The word that shall not be spoken. *New England Journal of Medicine* 2013 **369** 1777–1779. (https://doi.org/10.1056/NEJMp1309660)

2 Martínez-Momblán MA, Gómez C, Santos A, Porta N, Esteve J, Úbeda I, Halperin I, Campillo I, Guillaumet I, Webb SM *et al.* A specific nursing educational program in patients with cushing's syndrome. *Endocrine* 2015. (https://doi.org/10.1007/s12020-015-0737-0)

3 Gupta D, Lis CG & Rodeghier M. Can patient experience with service quality predict survival in colorectal cancer? *Journal for Journal of Healthcare Quality* 2013 **35** 37–43. (https://doi.org/10.1111/j.1945-1474.2012.00217.x)

4 Morel T & Cano SJ. Measuring what matters to rare disease patients – reflections on the work by the IRDiRC taskforce on patient-centered outcome measures. *Orphanet Journal of Rare Diseases* 2017 **12** 171. (https://doi.org/10.1186/s13023-017-0718-x)

5 Hoffman AR, Mathison T, Andrews D, Murray K, Kelepouris N & Fleseriu M. Adult growth hormone deficiency: diagnostic and treatment journeys from the patients' perspective. *Journal of the Endocrine Society* 2022 **6** bvac077. (https://doi.org/10.1210/jendso/bvac077)

6 Buhlman NW & Lee TH. When patient experience and employee engagement both improve, hospitals' ratings and profits climb. *Harvard Business Review*. Boston, MA, USA: Harvard Business Publishing, 2019. (available at https://hbr.org/2019/05/when-patient-experience-and-employee-engagement-both-improve-hospitals-ratings-and-profits-climb)

7 Webb SM, Kristensen J, Vitali D, van Klink S, van Beuzekom C, Santos A & Nordenström A. EndoERN patient survey on their perception of health care experience and of unmet needs for rare endocrine diseases. *Endocrine* 2021 **71** 569–577. (https://doi.org/10.1007/s12020-021-02625-0)

8 Bolz-Johnson M, Meek J & Hoogerbrugge N. "Patient Journeys": improving care by patient involvement. *European Journal of Human Genetics* 2020 **28** 141–143. (https://doi.org/10.1038/s41431-019-0555-6)

9 Aho-Glele U, Pomey MP, Gomes de Sousa MR & Bouabida K. An evidence-based tool (PE for PS) for healthcare managers to assess patient engagement for patient safety in healthcare organizations. *Patient Experience Journal* 2021 **8** 45–58. (https://doi.org/10.35680/2372-0247.1454)

10 Pomey MP, Ghadiri DP, Karazivan P, Fernandez N & Clavell N. Patients as partners: a qualitative study of patients' engagement in their health care. *PLOS ONE* 2015 **10** e0122499. (https://doi.org/10.1371/journal.pone.0122499)

11 Sibeoni J, Manolios E, Verneuil L, Chanson P & Revah-Levy A. Patients' perspectives on acromegaly diagnostic delay: a qualitative study. *European Journal of Endocrinology* 2019 **180** 339–352. (https://doi.org/10.1530/EJE-18-0925)

12 Shena N, Berniera T, Sequeiraa L, Straussa J, Silvera MP, Carter-Langforda A & Wiljera D. Understanding the patient privacy perspective on health information exchange: a systematic review. *International Journal of Medical Informatics* 2019 **125** 1–12. (https://doi.org/10.1016/j.ijmedinf.2019.01.014)
13 Street Jr RL, Gordon H & Haidet P. Physicians communication and perceptions of patients: is it how they look, how they talk, or is it just the doctor? *Social Science and Medicine* 2007 **65** 586–598. (https://doi.org/10.1016/j.socscimed.2007.03.036)
14 Santos A & Webb SM. Coping with Cushing's Disease: the Patients' Perspectives. *Cushing's Disease. An Often Misdiagnosed and Not So Rare Disorder*, pp. 169–185, ch. 10. Ed. ER Laws. Amsterdam, Netherlands: Elsevier, 2017. (https://doi.org/10.1016/B978-0-12-804340-0.00010-3)
15 Smaily H & Matar N. Laryngeal manifestations in acromegaly: a systematic review of the literature. *Acta Oto-Laryngologica Case Reports* 2020 **5** 57–63. (https://doi.org/10.1080/23772484.2020.1809421)
16 Yeoh P, Czuber-Dochan W, Aylwin S & Sturt J. Lived experience of people with adrenocortical carcinoma and associated adrenal insufficiency. *Endocrinology, Diabetes and Metabolism* 2022 **5** e341. (https://doi.org/10.1002/edm2.341)
17 Elyoussfi S. Understanding Patient Engagement: Exploring Factors around Utilisation of Screening and Surveillance Services in Individuals with or At Risk of Rare Endocrine Syndromes. *PhD Thesis*, Brunel University. (available at: https://bura.brunel.ac.uk/handle/2438/23505)
18 Ebbehoj A, Søndergaard E, Jepsen P, Stochholm K, Svane HML, Madsen M, Poulsen PL & Jørgensen JOL. The socioeconomic consequences of Cushing's syndrome: a nationwide cohort study. *Journal of Clinical Endocrinology and Metabolism* 2022 **107** 1–9. (https://doi.org/10.1210/clinem/dgac174)
19 Fønhus MS, Dalsbø TK, Johansen M, Frethei A, Skirbekk H & Flottorp SA. Patient-mediated interventions to improve professional practice. *Cochrane Database of Systematic Reviews* 2018 **9** CD012472. (https://doi.org/10.1002/14651858.CD012472.pub2)

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