Title: 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 (PREMs) and summarizes 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, for the benefit of those affected, but also for the entire health system.

Introduction
Knowledge on patient experience related to the healthcare systems is currently considered essential to pursue optimal quality of care (1). The concepts of PROMs (Patient-Reported Outcome Measures) and PREMs (Patient-Reported Experience Measures) are nowadays considered more and more important in health care evaluation; in fact, improved PREMs are shown to be associated with enhancement in the quality of healthcare, better financial outcome of health institutions and is even a predictor of survival (1-5). 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 every day needs. It enables 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 above.

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 on 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 health care professionals to improve patient outcome and experience, beyond the more technical issues, creating a positive patient-centred, empathic environment. Furthermore, a recent systematic review on 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 confusion and uncertainty of patients and their families. Waiting for a consultation can be very stressful, as well as having received conflicting information by 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, 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)

**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 (for example, 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 3 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 4 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).
Data of 6 patients from 4 countries were obtained (Tables 1 and 2). Diagnostic delay oscillated from 3 months to 15 years after 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 unrecognized by different health care 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 perceptible 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 a 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 for all. Clarifications of “scary” words like having a tumour in your head or requiring radiotherapy (associated to cancer) was stated as essential for the patients.
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, aging, weight gain or unhealthy life styles, for the majority, stress and impaired quality of life was 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, but 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 were 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 were 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 contacts with the health care system, is essential. Positively adapt to the life changes required is
important, and will help come to terms with the situation, while maintaining a satisfactory
daily quality of life.

Patient journey in Addison’s disease

Data of 6 patients from 4 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 recognized as such by the health care
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 salty food craving,
and worsening of all symptoms after minor health problem like a cold, an infection or
surgical procedure. Once recognized, however, all stated progressive physical and
psychological recovery as soon as glucocorticoid and mineralocorticoid substitution
therapy was initiated. Individualization 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, food and activity timetables, to optimize 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 health care 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 accessibility to parenteral hydrocortisone at hospital pharmacies, and patients plead to make it easier and more accessible.

**Patient journey in congenital adrenal hyperplasia**

Data of 7 patients from 3 countries were obtained (Tables 1 and 4); in one of the Italian reports, 2 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 to 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 new-born. 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 new-born 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 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 2 and half 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, was 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.

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 health care professionals and the patients’ close circles of family and friends was rather common. When relevant, identification through screening shortens the journey to diagnosis and prevents some of the initial 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, were included, a
vulnerable period when being aware of patients’ experiences and needs are of great importance.

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 it 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, 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.
Declaration of interest

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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


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Supplementary materials:

Supplementary Table 1: Patient journey (English version)

Supplementary Table 2: Patient journey translations into Danish, French, German, Italian, Spanish and Swedish
Table 1: Available translations and countries in which the Patient Journey template for different diseases was completed

<table>
<thead>
<tr>
<th>Diseases</th>
<th>Denmark</th>
<th>France</th>
<th>Germany</th>
<th>Italy</th>
<th>Lithuania (in English)</th>
<th>Spain</th>
<th>Sweden</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acromegaly</td>
<td>x</td>
<td>x</td>
<td></td>
<td></td>
<td>x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Addison's disease</td>
<td>x</td>
<td>x</td>
<td></td>
<td></td>
<td>x</td>
<td>x</td>
<td>x</td>
</tr>
<tr>
<td>Congenital adrenal hyperplasia</td>
<td>x</td>
<td>x</td>
<td>x</td>
<td></td>
<td>x</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Stage of Journey</td>
<td>Clinical Presentation/ Symptoms</td>
<td>Patient/Family Needs</td>
<td>Ideal Outcome/Support (how to address the needs)</td>
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<td></td>
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<tr>
<td>Pre-diagnostics</td>
<td>Typical Physical: Headache, carpal tunnel syndrome, sweating, menstrual irregularity, feeling tired with less strength, growth of hands, feet &amp; tongue (determining lispig), 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 &quot;something is wrong&quot; Psychological: Insomnia, lack of concentration, less memory, unreasonable nervousness, depression, fits of rage, irritability Less frequently described: &quot;Unexplained&quot; very large placenta, Laryngeal ventricular hernia, change in smile.</td>
<td>Two different scenarios:  - No suspicion of disease, problems thought to be related to stress, aging, weight gain or life-style 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 &amp; empathy by specialized multidisciplinary reference centers 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 center Information on radiotherapy, associated to &quot;cancer&quot; and scary  Earlier recognition by sleep centers, neurologists, ENT specialists, dentists, gynecologists, orthopedic surgeons, jewelers, shoe retailers, etc. Understanding that it's a chronic life-changing disease and learn to live with it, in a positive way</td>
<td>Listening to the patient by all involved in their health care (primary care, specialists consulted, nurses,) who complain they are not taken seriously despite knowing &quot;something is wrong&quot;. 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 organizing 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, acceptance and peace Access to psychiatrist or psychologist who understand the psychological aspects of the disease. Utility of questionnaires on QoL or different aspects of the disease which help the patient realize that they are validated and not crazy</td>
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</tbody>
</table>
### Table 3: Summary of Patient Journey data in Addison’s disease

<table>
<thead>
<tr>
<th>Stage of Journey</th>
<th>Timeline</th>
<th>Clinical Symptoms</th>
<th>Presentation/ (collective view)</th>
<th>Patient/Family Needs (collective view)</th>
<th>Ideal Outcome/Support (how to address the needs)</th>
</tr>
</thead>
</table>
| Pre-diagnosis    | 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 | No understanding by family, would appreciate more listening to the patients problems and concerns | 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. |  
| very symptomatic. After diagnoses and start of treatment great improvement although finding the exact dose required and its daily distribution may take some time | 10 years | 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 | Disease not recognized 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 |  
| 6 mo | 3-4 years | | | |  
| 1-2 years |  | | | |
Table 4: Data from Patient Journey' in congenital adrenal hyperplasia

<table>
<thead>
<tr>
<th>Stage of Journey</th>
<th>Timeline</th>
<th>Clinical Presentation/ Symptoms</th>
<th>Patient/Family Needs (collective view)</th>
<th>Ideal Outcome/Support (how to address the needs)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnosis</td>
<td>* 11 weeks of pregnancy 2 at birth 14 days postnatal 20 days postnatal ** 2.5 years of age</td>
<td>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</td>
<td>More attentive clinical staff/pediatrician to immediately recognize CAH, especially in the 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 favors 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</td>
<td>- Access to empathetic and knowledgeable HCP who immediately recognize 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 centers to optimize 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)</td>
</tr>
<tr>
<td>Screening diagnosis 8 days of age Boy with SW form</td>
<td></td>
<td>Irritable, difficult to comfort, but no electrolyte disturbance. Insufficient weight gain not identified before the screening result</td>
<td>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.</td>
<td>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.</td>
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</table>
TABLE 1: INFORMATION FOR THE PATIENTS AND TEMPLATE TO COLLECT DATA (ENGLISH VERSION)

PATIENT JOURNEY

Introduction

The European Reference Network for Rare Endocrine Diseases (EndoERN) aims at collecting information on how patients have experienced their disease. We as leaders of the work package 4 (WP4) focusing on patient care and quality of life, would like to ask you to participate in this pilot study, by reflecting your experience with your/your child’s disease. Our over-all aim is to improve patient outcome.

Description

Patient Journeys (PJ) represent the collective perspective on the burden of the disease and the needs of people with first-hand experience of living with a rare disease. They map the natural history and needs of patients along their life journey, through the patients’ or parents’ eyes.

The PJ can be used as a key tool to engage with the clinicians, to highlight the different needs of the patient community and identify how clinicians can respond to these needs. They can also be used for information to the patient community and to lay persons, like general practitioners and other healthcare workers.

The PJ shows in a comprehensive way the goals that are recognized by both patients and clinical experts. Therefore, it can be used by both these parties to explain the clinical pathway: professional experts can explain to newly identified patients how the clinical pathway generally looks like, whereas their patients can identify their specific needs within these pathways.

Suggested process

Please complete the table below including the different stages of the PJ that are relevant for you. Do not hesitate to adapt or modify the stages if needed but please do not delete or insert columns. Then, outline the clinical presentation and patient needs at each of the stages of the PJ and add a recommendation, or state what 'good care' and support would be, in the last column.

1. Complete the table template with your own experience, expectation, view and needs
2. Share this first version with your family and your patient community (e.g. members of your patient organizations and ask for feedback
3. Feel free to discuss the clinical presentation of your journey with your clinician / health care provider, if you wish.
4. Send back the filled in form by end January 2022.
5. WP4 will discuss these pilot PJ results with the Endo-ERN community to identify key common needs or priorities of patients with rare endocrine conditions, so the network can take positive action, for the benefit of the patients.
**INDICATE THE NAME OF THE RARE DISEASE/ CONDITION/ SYNDROME**

______________________________

**DATE**_________________            **COUNTRY OF RESIDENCE**________________

**INDICATE THE NUMBER OF PATIENTS/FAMILIES WHO CONTRIBUTED TO THIS PATIENT JOURNEY**

______________________________

“I have read and understood the attached “Informed Consent Privacy Statement related to collecting Information on Patient Journeys” and consent to the processing of the reported data as described therein”. [ ] (Please mark with an “x”)

<table>
<thead>
<tr>
<th>Stage of Journey Please adapt or modify stages if needed</th>
<th>Timeline</th>
<th>Clinical Symptoms</th>
<th>Presentation/ Patient/Family Needs (collective view)</th>
<th>Ideal Outcome/Support (how to address the needs)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pre-diagnosis</td>
<td></td>
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<td></td>
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<tr>
<td>First symptom</td>
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<tr>
<td>Diagnosis</td>
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<tr>
<td>First treatment</td>
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<tr>
<td>Surgery</td>
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<td></td>
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</tr>
<tr>
<td>Follow up care</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

..
Supplementary material: PATIENT JOURNEY TRANSLATIONS INTO DANISH, FRENCH, GERMAN, ITALIAN, SPANISH AND SWEDISH
PATIENTREJS

Introduktion

Det europæiske referencenetværk for sjældne endokrine sygdomme (Endo-ERN) har til formål at indsamle oplysninger om, hvordan patienter oplever deres sygdom. Som ledere af arbejdsgruppe 4 (WP4) inden for Endo-ERN, der fokuserer på patientbehandling og livskvalitet, vil vi gerne bede dig om at deltage i en pilotundersøgelse ved gennem en såkaldt PATIENTREJSE at give udtryk for dine erfaringer med din/dit barns sygdom på nedenstående skema. Vores overordnede mål er at medvirke til at forbedre patienternes behandling og livskvalitet.

Beskrivelse

Patientrejser (PR) repræsenterer det kollektive perspektiv på patienternes sygdomsbyrde og behov med førstehåndserfaring i at leve med en sjælden sygdom. De kortlægger patienternes historik og behov igennem deres sygdomsrejse set med patienternes eller forældrenes øjne.

PR kan bruges som et vigtigt redskab til at gå i dialog med hospitalerne, til at fremhæve patienternes forskellige behov og til at identificere, hvordan lægeverdenen kan imødekomme disse behov. De kan også bruges til at informere andre patienter og lægfolk, f.eks. praktiserende læger og andet sundhedspersonale.

PR viser på omfattende vis de mål, der er anerkendt af både patienter og kliniske eksperter. Derfor kan den bruges af begge parter til at forklare det kliniske forløb: faglige eksperter kan forklare nyligt diagnosticerede patienter, hvordan det kliniske forløb generelt ser ud, mens deres patienter kan identificere deres specifikke behov inden for disse forløb.

Foreslået proces


   1. Udfyld skabelonen med dine egne erfaringer, forventninger, synspunkter og behov
   2. Del denne første version med din familie og dit patientfællesskab (f.eks. medlemmer af din patientforening) og bed om feedback
   3. Du er velkommen til at drøfte den kliniske præsentation af din rejse med din behandlende læge/hospital, hvis du ønsker det.
   4. Send det udfyldte skema tilbage inden udgangen af marts 2022.
   5. WP4 vil herefter drøfte disse pilot-PR-resultater med Endo-ERN-netværket for at identificere vigtige fælles behov og prioriteter for patienter med sjældne endokrine lidelser, så netværket kan træffe de relevante foranstaltninger til gavn for patienterne.
ANGIV NAVNET PÅ DEN SJÆLDNE SYGDOM/TILSTAND/SYNDROM ________________

DATO ___________ BOPÆLSLAND ___________

ANGIV ANTALLET AF PATIENTER/FAMILIER, DER HAR BIDRAGET TIL DENNE PATIENTREJSE ________________

"Jeg har læst og forstået den vedlagte "Erklæring om informeret samtykke om beskyttelse af personlige oplysninger i forbindelse med indsamling af oplysninger om Patientrejser" og giver mit samtykke til behandling af de indberettede data som beskrevet heri". [ ] (Marker venligst med et "x")

<table>
<thead>
<tr>
<th>Stadie på rejsen</th>
<th>Tidslinje</th>
<th>Klinisk præsentation/symptomer</th>
<th>Patientens behov/familiens behov (kollektivt syn)</th>
<th>Det ideelle resultat/Den optimale støtte (hvordan imødekommes behovene)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tilpas eller modifier stadier om nødvendigt</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Pra-diagnose

Første symptomer

Diagnose

Første behandling

Evt. operation

Opfølgende behandling
LE PARCOURS DU PATIENT

Introduction
Le Réseau européen de référence pour les maladies endocriniennes rares (EndoERN) a pour but de collecter des informations sur la manière dont les patients ont vécu leur maladie. En tant que responsables du work package 4 (WP4) axé sur le soin des patients et la qualité de vie, nous aimerions vous demander de participer à cette étude pilote, reflétant votre expérience avec votre maladie ou celle de votre enfant. Notre objectif global est d'améliorer la prise en charge des patients.

Description
Le Parcours du Patient (PP) incarne une perspective collective qui se focalise sur le poids de la maladie et les besoins des patients ayants une expérience concrète de la vie avec une maladie rare. Les PP ont pour objectif de décrire l’histoire naturelle et les besoins des patients durant leur parcours de vie, à travers les yeux du patient ou de ses parents.

Les PP peuvent être utilisés comme un outil clé permettant aux cliniciens de l’ERN de mettre en lumière les différents besoins des patients et de comprendre comment répondre à ces besoins. Le PP peut aussi être utilisé pour l’information des patient ainsi que pour des personnes ayant de moindres connaissances dans les maladies endocriniennes rares, tels que des médecins généralistes ou autres professionnels de santé.

Le PP affiche de manière exhaustive les cibles reconnues à la fois par les patients et les experts cliniques. Par conséquent, il peut être utilisé par les deux parties pour expliquer le parcours clinique : des experts professionnels peuvent expliquer aux nouveaux patients à quoi peut ressembler leur parcours en général, tandis que les patients peuvent identifier les besoins spécifiques identifiés lors de ces visites.

Méthode suggérée
Veuillez compléter le tableau ci-dessous comportant les différentes étapes (les plus intéressantes selon vous) du parcours du patient. N’hésitez pas à adapter ou à modifier les étapes si besoin mais n’insérez ni n’effacez pas de colonnes s’il vous plaît. Ensuite, exposez la présentation clinique et les besoins des patients à chaque étape du PP et ajoutez une recommandation ou décrivez ce que serait le “bon soin” dans la dernière colonne.

1. Complétez le tableau en vous basant sur vos propres expériences/attentes/avis/besoins.
2. Partagez, si vous voulez, cette première version avec votre famille et communautés de patients (membres d’associations de patients) et demandez des retours.
3. Si vous voulez, partagez la présentation clinique de votre parcours avec votre clinicien (optionnel).
4. Retournez la fiche avant la fin de Janvier 2022.
5. Le WP4 discutera de ces résultats pilotes de RP avec la communauté Endo-ERN pour identifier les principaux besoins ou priorités communs des patients atteints de maladies endocriniennes rares, afin que le réseau puisse prendre des mesures positives au profit des patients.
**INDIQUER LE NOM DE LA MALADIE / ÉTAT / SYNDROME MINORITAIRE**

_____________________________________________________

**DATE___________ PAYS___________________**

**INDIQUER LE NOMBRE DE PATIENTS/FAMILLES CONTRIBUANT A CET ECRITURE**

_____________________________________________________

J'ai lu et compris la « Déclaration de confidentialité du consentement éclairé relative à la collecte d'informations sur les voyages des patients » et je consens au traitement des données signalées tel qu'il y est décrit. « [] (Veuillez cocher un « x »)

<table>
<thead>
<tr>
<th>Stade du parcours</th>
<th>Chronologie</th>
<th>Présentation clinique/Symptômes</th>
<th>Besoins du patient/de la famille (vision collective)</th>
<th>Résultat/soutien idéal (comment répondre aux besoins)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Modifiez les stades si besoin</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

- Pre-diagnostic
- Premiers symptômes
- Diagnostic
- Premier traitement
- Chirurgie
- Soins de suivi
PATIENTENREISE

Einführung
Das Europäische Referenznetzwerk für seltene endokrine Erkrankungen (EndoERN) hat sich zum Ziel gesetzt, Informationen darüber zu sammeln, wie Patienten ihre Krankheit erlebt haben. Als Leiter des Arbeitspakets 4 (WP4), das sich mit der Patientenversorgung und der Lebensqualität befasst, möchten wir Sie bitten, an dieser Pilotstudie teilzunehmen, indem Sie Ihre Erfahrungen mit Ihrer Krankheit, der Ihres Kindes oder Ihrer Familie schildern. Unser übergeordnetes Ziel ist es, die Ergebnisse für die Patienten zu verbessern.

Beschreibung

Vorgeschlagener Prozess
Füllen Sie bitte die nachstehende Tabelle aus und geben Sie die verschiedenen Phasen des PJ an, die für Sie relevant sind. Zögern Sie nicht, die Stadien bei Bedarf anzupassen oder zu ändern, aber löschen oder fügen Sie bitte keine Spalten ein. Skizzieren Sie dann das klinische Bild und die Bedürfnisse des Patienten in jeder Phase des PJ und fügen Sie in der letzten Spalte eine Empfehlung hinzu oder geben Sie an, was eine "gute Pflege" und Unterstützung wäre.
1. Füllen Sie die Tabellenvorlage mit Ihren eigenen Erfahrungen, Erwartungen, Ansichten und Bedürfnissen aus.
3. Diskutieren Sie die klinische Darstellung Ihrer Reise mit Ihrem Arzt / Gesundheitsdienstleister, wenn Sie dies wünschen.
4. Schicken Sie das ausgefüllte Formular bis Ende Januar 2022 zurück.
5. WP4 wird die Ergebnisse des PJ-Pilotprojekts mit der Endo-ERN-Gemeinschaft diskutieren, um die wichtigsten gemeinsamen Bedürfnisse oder Prioritäten von Patienten mit seltenen endokrinen Erkrankungen zu ermitteln, damit das Netzwerk positive Maßnahmen zum Wohle der Patienten ergreifen kann.
GEBEN SIE DEN NAMEN DER SELTENEN KRANKHEIT/DES LEIDENS/DES SYNDROMS AN ___________
DATUM_________ LAND ________ DES _______ WOHNSITZES___________
GEBEN SIE DIE ANZAHL DER PATIENTEN/FAMILIEN AN, DIE ZU DIESER PATIENTENREISE BEIGETRAGEN HABEN_______

"Ich habe die beigefügte "Datenschutzerklärung zur Erfassung von Informationen über Patientenreisen" gelesen und verstanden und stimme der darin beschriebenen Verarbeitung der gemeldeten Daten zu". X (Bitte mit einem "x" markieren)

<table>
<thead>
<tr>
<th>Stadium der PJ (evtl. anpassen)</th>
<th>Zeitablauf</th>
<th>Klinische Darstellung / Symptome</th>
<th>Bedürfnisse des Patienten/ der Familie (zusammengefasste Darstellung)</th>
<th>Ideales Ergebnis/ Ideale Unterstützung der Bedürfnisse</th>
</tr>
</thead>
<tbody>
<tr>
<td>Vor-Diagnose</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Erstes Symptom</td>
<td></td>
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<tr>
<td>Diagnose</td>
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<tr>
<td>Erste Behandlung</td>
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<tr>
<td>Eventuelle Operation</td>
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<tr>
<td>Nachsorge</td>
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</tbody>
</table>

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IL PERCORSO DEL PAZIENTE

Introduzione
La rete di riferimento europea per le malattie endocrine rare (EndoERN) mira a raccogliere informazioni su come i pazienti vivono la propria malattia. Come leaders del Working Package 4 (WP4) che si concentra su “Punto di vista del paziente e qualità della vita”, vorremmo chiederti di partecipare a questo studio pilota, riflettendo sulla tua esperienza con la malattia. Il nostro obiettivo generale è quello di migliorare la qualità della vita per il paziente.

Descrizione
I Patient Journeys (PJ) rappresentano la prospettiva collettiva sul peso della malattia e i bisogni delle persone con la propria esperienza nel vivere con una malattia rara. Essi mappano la storia naturale e i bisogni dei pazienti lungo il loro percorso di vita, attraverso gli occhi dei pazienti o dei familiari.

I PJ possono essere usati come uno strumento chiave per coinvolgere i clinici, per evidenziare i diversi bisogni della comunità dei pazienti e identificare come i clinici possono rispondere a questi bisogni. Possono anche essere usati per informare la comunità dei pazienti e operatori non esperti, come i medici di base e altri operatori sanitari.

Il PJ mostra in modo completo gli obiettivi che sono riconosciuti sia dai pazienti che dagli esperti clinici. Pertanto, può essere usato da entrambe queste parti per spiegare il percorso clinico: gli esperti professionali possono spiegare ai pazienti appena identificati come appare il percorso clinico in generale, mentre i loro pazienti possono identificare i loro bisogni specifici all'interno di questi percorsi.

Processo suggerito
Completate la tabella qui sotto includendo le diverse fasi del PJ che sono rilevanti per voi. Non esitate ad adattare o modificare le fasi se necessario, ma per favore non cancellate o inserite colonne. Poi, delineate la presentazione clinica e i bisogni del paziente in ciascuna delle fasi del PJ e aggiungete una raccomandazione, o dichiarate quali sarebbero le "buone pratiche" e il supporto, nell'ultima colonna.

1. Completate il modello della tabella con la vostra esperienza, aspettativa, visione e necessità
2. Condividi questa prima versione con la tua famiglia e la tua comunità di pazienti (ad esempio i membri della tua organizzazione di pazienti e chiedi un feedback
4. Rispedisce il modulo compilato entro la fine di gennaio 2022.
5. Il WP4 discuterà questi risultati del PJ pilota con la comunità Endo-ERN per identificare le principali esigenze o priorità comuni dei pazienti con condizioni endocrine rare, in modo che la rete possa intraprendere azioni positive, a beneficio dei pazienti.
**INDICARE IL NOME DELLA MALATTIA/CONDIZIONE/SINDROME RARA**

__________________________

**DATA__________ PAESE DI RESIDENZA__________**

**INDICARE IL NUMERO DI PAZIENTI/FAMIGLIE CHE HANNO CONTRIBUITO A QUESTO “VIAGGIO DEL PAZIENTE”**

__________________________

"Ho letto e compreso l'allegata "Informativa sulla raccolta dei dati per la partecipazione al Patient Journey" e acconsento al trattamento dei dati riportati come ivi descritto". [x] (Segnare con una "x")

<table>
<thead>
<tr>
<th>Fase del viaggio</th>
<th>Linea Temporale</th>
<th>Presentazione clinica/sintomi</th>
<th>Bisogni del paziente/famiglia (punto di vista collettivo)</th>
<th>Risultato ideale/sostegno (come affrontare i bisogni)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prima della diagnosi</td>
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<tr>
<td>Primi sintomi</td>
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<tr>
<td>Diagnosi</td>
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<tr>
<td>Prime terapie</td>
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<tr>
<td>Interventi chirurgici</td>
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<tr>
<td>Cure e follow up</td>
<td></td>
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</tr>
</tbody>
</table>
RECORRIDO DEL PACIENTE

Introducción
La Red europea de referencia para enfermedades endocrinas raras (EndoERN) tiene como objetivo recopilar información sobre cómo los pacientes han experimentado su enfermedad. Como líderes del paquete de trabajo 4 (work Packaged 4 -WP4-) centrado en la atención al paciente y la calidad de vida, nos gustaría pedirle que participe en este estudio piloto, reflejando su experiencia con su enfermedad o la de su hijo. Nuestro objetivo general es mejorar el resultado del paciente.

Descripción
Los recorridos del paciente (RP) representan la perspectiva colectiva sobre la carga de la enfermedad y las necesidades de las personas con experiencia de primera mano de vivir con una enfermedad rara o minoritaria. Describen la historia natural y las necesidades de los pacientes a lo largo de su trayectoria vital, desde el punto de vista del paciente o de sus padres.

Estos recorridos (en inglés “journey” o viaje) de los pacientes se pueden utilizar como una herramienta clave para interactuar con los responsables clínicos a fin de resaltar las diferentes necesidades de la comunidad de pacientes e identificar cómo la ERN puede responder a estas necesidades. También se pueden utilizar para facilitar información para el colectivo de pacientes y para otros colectivos como médicos generales y otros trabajadores de la salud.

El RP muestra de manera integral los objetivos que son reconocidos tanto por pacientes como por expertos clínicos. Por lo tanto, ambas partes pueden utilizarlo para explicar el recorrido clínico: los expertos profesionales pueden explicar a los pacientes recién identificados cómo se podrá ser su recorrido en general, mientras que los pacientes pueden identificar necesidades específicas identificadas en estos recorridos.

Proceso sugerido
Complete la tabla adjunta, incluyendo las diferentes etapas del recorrido del paciente que sean relevantes para usted. No dude en adaptar o modificar las etapas si es necesario, pero no elimine ni inserte columnas. Luego, describa la presentación clínica y las necesidades del paciente en cada una de las etapas del recorrido del paciente y agregue una recomendación o indique lo que considera sería un "buen cuidado" y buen soporte en la última columna.

1. Complete la plantilla de la tabla con su propia experiencia, expectativas, punto de vista y necesidades.
2. Comparta (si lo desea) esta primera versión con familia y comunidad de pacientes (miembros de sus organizaciones de pacientes, Federación Europea, etc.)
3. Si lo desea puede discutir la presentación clínica de su recorrido con su sanitario de referencia,
4. Remita la tabla completada como muy tarde a final de enero 2022
5. WP4 discutirá estos resultados piloto de RP con la comunidad Endo-ERN para identificar necesidades o prioridades comunes clave de los pacientes con afecciones endocrinas raras, para que la red pueda tomar medidas positivas en beneficio de los pacientes.
He leído y comprendo la "Declaración de privacidad de consentimiento informado relacionada con la recopilación de información sobre los viajes de los pacientes" y doy mi consentimiento para el procesamiento de los datos notificados como se describe en la misma. [ ] (Marque con una "x")

<table>
<thead>
<tr>
<th>Etapa del recorrido</th>
<th>Cronología</th>
<th>Presentación Clínica/ Síntomas</th>
<th>Necesidades del paciente/Familia (vista colectiva)</th>
<th>Resultado Ideal /Soporte (cómo abordar las necesidades)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pre-diagnóstico</td>
<td></td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Primer síntoma</td>
<td></td>
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<tr>
<td>Diagnóstico</td>
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<td>Primer tratamiento</td>
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<tr>
<td>Cirugía</td>
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<td></td>
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<tr>
<td>Seguimiento</td>
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</tr>
</tbody>
</table>
**INTRODUKTION**

EndoERN, (European Reference Network for Rare Endocrine Diseases- ett stort europeiskt nätverk för ovanliga endokrina sjukdomar) vill samla information om hur patienter upplever sin sjukdom. Vi som skriver till dig nu, leder en grupp inom nätverket (WP4) som fokuserar på patientens vård och livskvalitet. Vi vill fråga om du vill delta i en pilotstudie och beskriva dina erfarenheter av din/ ditt barns sjukdom. Vårt övergripande mål är att förbättra för patienterna.

**Beskrivning**

Patientens resa (PJ) är tänkt att beskriva det gemensamma perspektivet på sjukdomsbördan och behoven hos personer med förstahandserfarenhet, det vill säga erfarenhet av att leva med en sällsynt sjukdom. En kartläggning av naturalförloppet och behoven längs personernas livsresa, genom patienternas eller föräldrarnas ögon.

PJ kan användas som ett verktyg för att underlätta samarbete mellan patienter och kliniska experter för att lyfta fram de olika behov som patientgruppen har och identifiera hur kliniker kan svara upp mot dessa behov. PJ kan också användas för information till patientgruppen, allmänheten och till allmänläkare och annan vårdpersonal.

PJ visar på ett övergripande sätt vilka mål som identifieras av både patienter och kliniska experter. Den kan därför användas av båda parter för att förklara det kliniska förloppet: professionella experter kan förklara för nydiagnostiserade patienter hur förloppet i stort ser ut, medan patienterna kan identifiera de specifika behov de har inom det kliniska förloppet.

**Förslag till arbetsprocess**

Fyll i tabellmallen nedan, inklusive de olika stadierna av patientens resa som är relevanta för dig. Tveka inte att anpassa eller modifiera de olika stadierna om det behövs, men ta inte bort eller infoga kolumner. Beskriv sedan den kliniska presentationen, symtomen och patientbehoven i varje steg av patientens resa och lägg till en rekommendation eller ange vad god vård och stöd skulle vara i den sista kolumnen.

1. Fyll i tabellmallen med utgångspunkt från din egen erfarenhet, förväntningar, åsikter och behov. Beskriv det som är relevant för dig.
2. Visa denna första version för dina närmaste och sedan för en större grupp patienter (medlemmar i din patientorganisation, EU, etc) och be om synpunkter.
3. Diskutera den kliniska presentationen av din resa med din läkare om du vill
4. Skicka tillbaka den ifyllda tabellen före sista mars (2022-03-31)
5. Vi i WP4 kommer att diskutera PJ resultat med EndoERN för att identifiera gemensamma behov eller prioriteringar som patienter med ovanliga endokrina sjukdomar har för att nätverket ska kunna arbeta för förbättringar för patienterna.

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VILKEN OVANLIG ENDOKRIN SJUKDOM BESKRIVS___________________

DATUM______________   LAND______________

ANTAL PATIENTER/FAMILJER SOM HAR BIDRAGIT TILL DEN HÄR "PATIENT RESAN"___________________

"Jag har läst och förstått den bifogade informationen angående insamling av data och deltagande i Patientens Resa och samtycker till att data insamlas och bearbetas såsom beskrivet.”

[   ] (Vänligen markera med ett “X”)

<table>
<thead>
<tr>
<th>Stadium av ”resan”</th>
<th>Timeline</th>
<th>Klinska Symptom</th>
<th>Presentation</th>
<th>Patient/Familjens behov (tillsammans)</th>
<th>Ideal situation/Support (hur kan behoven mötas)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pre-diagnos/före diagnos</td>
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