

Section 1

Introduction and methods

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Introduction

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease.

A total of 407 participants with rare diseases or carers to people with rare diseases were recruited into this PEEK study. There were 392 that completed both parts of the study, 5 that completed or partially completed online questionnaire only and 10 participants that completed the interview only.

Personal Experience, Expectations and Knowledge (PEEK)

Patient Experience, Expectations and Knowledge (PEEK) is a research program developed by the Centre for Community-Driven Research (CCDR). The aim of PEEK is to conduct patient experience studies across several disease areas using a protocol that will allow for comparisons over time (both quantitative and qualitative components). PEEK studies give us a clear picture and historical record of what it is like to be a patient at a given point in time, and by asking patients about their expectations, PEEK studies give us a way forward to support patients and their families with treatments, information and care.

The research protocol used in PEEK studies is independently driven by CCDR. PEEK studies include a quantitative and qualitative component. The quantitative component is based on a series of validated tools. The qualitative component is the result of two years of protocol testing by CCDR to develop a structured interview that solicits patient experience data and provides patients with the opportunity to provide advice on what they would like to see in relation to future treatment, information and care. The structured interview has also been designed so that the outcomes of PEEK studies can inform policy, research, care, information, supportive care services and advocacy efforts.

Position of this study

A search was conducted in Pubmed (August 8, 2022) to identify studies of rare diseases with that described patient experience conducted in the past five years in Australia, and updated on January 4th 2023. The term "Rare disease" was searched in any field, and it is noted that not all rare diseases studies will be included using this search term, and the difficulty in searching using individual disease names. Interventional studies, meta-analysis studies, studies conducted in developing countries, and studies of less than five participants were excluded.

There were 201 studies identified, 52 studies used interviews, 30 studies used focus groups or other qualitative methods and 138 studies used questionnaires.

PEEK is largest study of rare diseases conducted in an Australian population with a total of 402 participants with rare diseases or carers to people with rare diseases were recruited into the study. There were 391 that completed or partially completed online questionnaires and 402 participants that were interviewed.

Introduction

In Australia, a disease is considered rare if it affects less than 5 in 10,000 people. There are more than 7,000 rare diseases that are life threatening or chronically debilitating. Around 8% of Australians (2 million people) live with a rare disease¹.

A total of 407 participants with rare diseases or carers to people with rare diseases were recruited into this PEEK study. There were 392 that completed both parts of the study, 5 that completed or partially completed online questionnaire only and 10 participants that completed the interview only.

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Participants

To be eligible for the study, participants needed to have been diagnosed with a rare disease or be a carer to a person with a rare disease, have experienced the healthcare system in Australia, be 18 years of age or older, be able to speak English, and be able to give consent to participate in the study.

Ethics

Ethics approval for this study was granted (as a low or negligible risk research study) by the Centre for Community-Driven Research Ethics Committee (Reference CS_Q4_03).

Data collection

Data for the online questionnaire was collected using Zoho Survey (Zoho Corporation Pvt. Ltd. Pleasanton, California, USA, www.zoho.com/survey).

There were five researchers who conducted telephone interviews and used standardised prompts throughout the interview. The interviews were recorded and transcribed verbatim. Identifying names and locations were not included in the transcript. All transcripts were checked against the original recording for quality assurance.

Online questionnaire (quantitative)

The online questionnaire consisted of the 36-Item Short Form Health Survey (SF36) (RAND Health)², a modified Cancer Care Coordination Questionnaire for Patients (CCCQ)³, the Short Fear of Progression Questionnaire (FOP12)⁴, and the Partners in Health version 2 (PIH)⁵. In addition, investigator derived questions about demographics, diagnosis, treatment received and future treatment decisions making were included.

Structured Interview (qualitative)

Interviews were conducted via telephone by registered nurses who were trained in qualitative research. The first set of interview questions guided the patient through their whole experience from when symptoms were noticed up to the present day.

Questionnaire analysis

Statistical analysis was conducted using R included in the packages “car”, “dplyr” and “ggplot2” (R 3.3.3 GUI 1.69 Mavericks build (7328)). The aim of the statistical analysis of the SF36, CCCQ, FOP12, and PIH responses was to identify variations by condition, type or participant (person with condition or carer), gender, age, education, location of residence, education status and socio-economic status. Scales and subscales were calculated according to reported instructions²⁻⁵.

The Location of participants was evaluated by postcode using the Australian Statistical Geography Maps (ASGS) Remoteness areas accessed from the Australian Bureau of Statistics⁶.

The level of socio-economic status of participants was evaluated by postcode using the Socio-economic Indexes for Areas (SEIFA) accessed from the Australian Bureau of Statistics⁷.

For comparisons by condition and age, a one-way analysis of variance (ANOVA) analysis was conducted. A Tukey HSD test was used post-hoc to identify the source of any differences identified in the one-way ANOVA test. Where the assumptions for the one-way ANOVA were not met, a Kruskal-Wallis rank sum test on care was conducted with post-hoc pairwise comparisons using Wilcoxon rank sum test. When the assumption of equal variances were not met, a Welch one-way test was used with post-hoc pairwise t-tests with no assumption of equal variances.

For all other comparisons between groups, a two-sample t-test was used when assumptions for normality and variance were met, or when assumptions were not met, a Wilcoxon rank sum test with continuity correction was used. Questions where participants were asked to rank preferences were analysed using weighted averages. Weights were applied in reverse, the most preferred option was given the largest weight equal to the number of options, the least preferred option was given the lowest weight of 1.

Structured interviews analysis

A content analysis was conducted using conventional analysis to identify major themes from structured interviews. Text from the interviews were read line-by-line by the lead researcher and then imported into the custom PEEK analysis database. Each question within the interview was individually analysed. Initial categories and definitions were identified and registered in custom PEEK analysis database. The minimum coded unit was a sentence with paragraphs and phrases coded as a unit.

A second researcher verified the codes and definitions, and the text was coded until full agreement was reached using the process of consensual validation. Where a theme occurred less than 5 times it was not included in the study results, unless this result demonstrated a significant gap or unexpected result.

Data analysis and final reporting was completed in November 2023.

Position of this study

A search was conducted in Pubmed (August 8, 2022) to identify studies of rare diseases with that described patient experience conducted in the past five years in Australia, and updated on January 4th 2023 (Table 1.1). The term "Rare disease" was searched in any field, and it is noted that not all rare diseases studies will be included using this search term, and the difficulty in searching using individual disease names. Interventional studies, meta-analysis studies, studies conducted in developing countries, and studies of less than five participants were excluded. All studies identified are listed in Table 1.1 alongside the country where the study was conducted, the number of participants by data collection method, and the broad focus of the study.

There were 201 studies identified, 52 studies used interviews, 30 studies used focus groups or other qualitative methods and 138 studies used questionnaires.

The most common disease areas covered were endocrine, nutritional or metabolic diseases (n=43), developmental anomalies (n=35), diseases of the immune system (n=26), groups of diseases (n=23), diseases of the nervous system (n=17), neoplasms (n=13), and diseases of the respiratory system (n=10).

There were 4 studies conducted completely with an Australian population, one study of 25 participants that took part in focus groups, with 190 participants completing surveys about expectations of future care and treatment⁸, one study of 20 participants that took part in focus groups also focused on future treatments and expectations⁹, one study of 184 participants who completed surveys about decision making¹⁰, and one study of 50 participants that completed surveys about health related quality of life¹¹.

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Table 1.1: PEEK position

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Babac (2018) ^{12,13}	Groups of diseases	Germany	68	0	0	Decision making
Guffon (2022) ¹⁴	Endocrine, nutritional or metabolic diseases	France	55	0	0	unmet needs
Le Hénaff (2020) ¹⁵	Diseases of the skin	France	50	0	0	Quality of life
Capella-Peris (2020) ¹⁶	Diseases of the nervous system	USA	47	0	0	Symptoms and side effects
Hoffmann-Vold (2021) ¹⁷	Diseases of the immune system	Multinational	47	0	0	Expectations
Moffatt (2019) ¹⁸⁻²¹	Diseases of the circulatory system	Multinational	46	40	1203	Self-management
Kerr (2023) ²²	Multiple rare diseases	USA	45	0	0	Navigating healthcare
Pokrzywinski (2021) ²³	Endocrine, nutritional or metabolic diseases	Multinational	44	0	0	Quality of life
Peter (2022) ²⁴	Groups of diseases	UK	38	65	0	Symptoms and diagnosis
Harrington (2019) ²⁵	Endocrine, nutritional or metabolic diseases	USA	32	0	0	Symptoms and diagnosis
Eichler (2022) ²⁶	Endocrine, nutritional or metabolic diseases	Multi-national	31	0	0	Symptoms
Young (2022) ²⁷	Undiagnosed diseases	USA	30	0	0	Participation in research
Munro (2022) ²⁸	Groups of diseases	USA	29	0	29	Quality of life
Pompilus (2021) ²⁹	Neoplasms	USA	27	0	0	Quality of life
Bingaman (2022) ³⁰	Metabolic disorders	Multi-national	26	13	0	Symptoms
Kutsa (2022) ³¹	Diseases of the immune system	USA	26	0	0	Parental coping
Vu Minh Arnell (2019) ³²	Developmental anomalies	Sweden	25	0	0	Quality of life
Pascoal (2022) ³³	Endocrine, nutritional or metabolic diseases	Portugal	23	0	23	Symptoms
Khanna (2020) ³⁴	Diseases of the immune system	Multinational	23	0	0	Symptoms and diagnosis
Lanar (2022) ³⁵	Endocrine, nutritional or metabolic diseases	Multinational	22	0	0	Quality of life
Gregersen (2022) ³⁶	Neoplasms	Denmark	22	0	0	Symptoms and diagnosis
Gumuchian (2018) ³⁷	Diseases of the skin	Canada	22	0	0	Quality of life
Nguyen (2022) ³⁸	Endocrine, nutritional or metabolic diseases	France	21	0	0	Quality of life
Iyer (2020) ³⁹	Diseases of the nervous system	USA	20	0	0	Self-management
Lewis (2020) ⁴⁰	Groups of diseases	UK	20	0	0	Symptoms and diagnosis
Hunter (2019) ⁴¹	Diseases of the nervous system	USA	20	0	0	Quality of life
de Dios García-Díaz (2022) ⁴²	Endocrine, nutritional or metabolic diseases	Spain	20	0	0	Expectations
Güeita-Rodríguez (2020) ⁴³	Developmental anomalies	Spain	19	31	0	Care and support
Merker (2021) ⁴⁴	Developmental anomalies	USA	18	0	0	Symptoms and diagnosis
Powell (2022) ⁴⁵	Diseases of the nervous system	UK	18	0	0	Quality of life
Lyn (2020) ⁴⁶	Endocrine, nutritional or metabolic diseases	USA	17	19	0	Quality of life
Olivotto (2022) ⁴⁷	Diseases of the nervous system	Italy	16	0	16	Health-related quality of life
Plackowski (2022) ⁴⁸	Multiple rare diseases	USA	16	9	0	Self advocacy/lived experience
Granero-Molina (2020) ⁴⁹	Diseases of the immune system	Spain	16	6	0	Symptoms and diagnosis
Baumbusch (2018) ⁵⁰	Groups of diseases	Canada	16	0	0	Care and support
Jimenez-Moreno (2021) ⁵¹	Diseases of the nervous system	UK	15	52	0	Decision making

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Simpson (2021) ⁵²	Groups of diseases	UK	15	0	0	Care and support
Stanarević Katavić (2019) ⁵³	Groups of diseases	Croatia	15	0	0	Self-management
Livermore (2019) ⁵⁴	Diseases of the immune system	UK	15	0	0	Quality of life
Grimstvedt (2021) ⁵⁵	Diseases of the nervous system	Norway	14	0	0	Treatment and management
Lee (2022) ⁵⁶	Groups of diseases	Canada	14	0	0	Symptoms and diagnosis
Deutch (2021) ⁵⁷	Undiagnosed	USA	14	0	0	Care and support
Wheeden (2022) ⁵⁸	Endocrine, nutritional or metabolic diseases	Multinational	14	0	0	Symptoms and side effects
van Dongen (2022) ⁵⁹	Neoplasms	Netherlands	14	0	0	Condition management
Hausmann (2018) ⁶⁰	Diseases of the immune system	USA	12	0	0	Symptoms and diagnosis
Smits (2022) ⁶¹	Groups of diseases	Netherlands	12	0	0	Care and support
Ford (2019) ⁶²	Diseases of the nervous system	UK	11	0	71	Symptoms and side effects
Honingh (2022) ⁶³	Endocrine, nutritional or metabolic diseases	Netherlands	11	0	0	Care and support
Adams (2018) ⁶⁴	Diseases of the skin	UK	11	0	0	Quality of life
McCausland (2018) ⁶⁵	Endocrine, nutritional or metabolic diseases	USA	10	0	341	Symptoms and diagnosis
Ragan (2021) ⁶⁶	Diseases of the digestive system	Canada	10	0	0	Self-management
Gaasterland (2019) ⁶⁷	Groups of diseases	Netherlands	10	0	0	Expectations
McLaughlin (2022) ⁶⁸	Diseases of the blood or blood-forming organs	UK	3	11	0	Symptoms
Eskes (2022) ⁶⁹	Endocrine, nutritional or metabolic diseases		2	37	0	Treatment
Erbis (2018) ⁷⁰	Diseases of the immune system	Germany	0	78	40	Expectations
Swezey (2019) ⁷¹	Developmental anomalies	Multinational	0	75	0	Expectations
Dwyer (2022) ⁷²	Endocrine, nutritional or metabolic diseases	USA	0	58	0	Symptoms and diagnosis
Knoppers (2022) ⁷³	Diseases of the respiratory system	Canada	0	40	0	Expectations
Velvin (2021) ⁷⁴	Developmental anomalies	Norway	0	36	0	Treatment and management
Milette (2019) ⁷⁵	Diseases of the skin	Canada	0	34	0	Quality of life
Tikellis (2021) ⁸	Diseases of the respiratory system	Australia	0	25	190	Expectations
Houdayer (2019) ⁷⁶	Undiagnosed	France	0	21	0	Symptoms and diagnosis
Long (2021) ⁹	Endocrine, nutritional or metabolic diseases	Australia	0	20	0	Expectations
Milette (2020) ⁷⁷	Diseases of the skin	Canada	0	19	0	Care and support
Uhlenbusch (2019) ⁷⁸	Groups of diseases	Germany	0	18	0	Quality of life
Casassa (2021) ⁷⁹	Developmental anomalies	France	0	16	0	Quality of life
Bate (2019) ⁸⁰	Neoplasms	UK	0	10	18	Decision making
Younger (2022) ⁸¹	Developmental anomalies	USA	0	10	0	multi-specialist clinic
Verger (2021) ⁸²	Groups of diseases	Spain	0	9	0	Care and support
Quinn (2020) ⁸³	Groups of diseases	Multinational	0	8	251	Self-management
Kocher (2021) ⁸⁴	Diseases of the immune system	Switzerland	0	5	101	Self-management
Mälstam (2018) ⁸⁵	Endocrine, nutritional or metabolic diseases	Sweden	0	5	0	Quality of life
Danvers (2022) ⁸⁶	Diseases of the immune system	France	0	*	0	Online support
Strobel (2022) ⁸⁷	Diseases of the immune system	Multi-national	0	*	0	Online support
Schwartz (2017) ⁸⁸	Groups of diseases	USA	0	0	3324	Quality of life

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Tse (2021) ⁸⁹	Diseases of the immune system	Multinational	0	0	3233	Expectations
Doser (2022) ⁹⁰	Developmental anomalies	Denmark	0	0	2467	Health-related quality of life
McMillan (2021) ⁹¹	Diseases of the nervous system	Canada	0	0	1927	Treatment and management
Catto (2021) ⁹²	Neoplasms	UK	0	0	1796	Health-related quality of life
Spencer-Tansley (2022) ⁹³	Groups of diseases	UK	0	0	1795	Treatment and management
de Graaf (2021) ⁹⁴	Endocrine, nutritional or metabolic diseases	Multinational	0	0	1378	Expectations
Bogart (2017) ⁹⁵	Groups of diseases	USA	0	0	1218	Health-related quality of life
Wunsch (2022) ⁹⁶	Diseases of the immune system	Multi-national	0	0	1178	Health-related quality of life
Szczepura (2018) ⁹⁷	Groups of diseases	UK	0	0	1158	Symptoms and diagnosis
Bogart (2022) ⁹⁸	Groups of diseases	USA	0	0	1118	Care and support
Eichler (2022) ^{99,100}	Neoplasms	Germany	0	0	1113	Health-related quality of life
Springer (2020) ¹⁰¹	Diseases of the immune system	USA	0	0	926	Symptoms and side effects
Thyen (2018) ¹⁰²	Developmental anomalies	Multinational	0	0	848	Health-related quality of life
Garrido-Esteba (2022) ¹⁰³	Injury, poisoning or certain other consequences of external causes	Spain	0	0	828	Health-related quality of life
Azar (2018) ¹⁰⁴	Diseases of the skin	Multinational	0	0	752	Treatment and management
Lancaster (2022) ¹⁰⁵	Diseases of the respiratory system	Multinational	0	0	739	Care and support
Cottin (2022) ¹⁰⁶	Diseases of the respiratory system	France	0	0	724	Symptoms and side effects
Hiremath (2018) ¹⁰⁷	Diseases of the digestive system	USA	0	0	632	Care and support
Duret (2020) ¹⁰⁸	Diseases of the immune system	France	0	0	632	Symptoms and side effects
Chu (2022) ¹⁰⁹	Diseases of the immune system	USA	0	0	629	Health-related quality of life
Damy (2022) ¹¹⁰	Endocrine, nutritional or metabolic diseases	France	0	0	603	Symptoms and diagnosis
Webb (2021) ¹¹¹	Endocrine, nutritional or metabolic diseases	Multinational	0	0	598	Care and support
Montali (2021) ¹¹²	Diseases of the digestive system	Multinational	0	0	569	Health-related quality of life
Kimura (2022) ¹¹³	Certain infectious or parasitic diseases	Japan	0	0	538	Health-related quality of life
Hanisch (2018) ^{114,115}	Groups of diseases	Germany	0	0	484	Health-related quality of life
Tsai (2021) ¹¹⁶	Developmental anomalies	USA	0	0	468	Expectations
Al Mukaddam (2022) ¹¹⁷	Diseases of the musculoskeletal system or connective tissue	Multi-national	0	0	463	Health-related quality of life
Kreuter (2019) ¹¹⁸	Diseases of the respiratory system	Germany	0	0	424	Health-related quality of life
Park (2019) ¹¹⁹	Diseases of the immune system	Korea	0	0	360	Health-related quality of life
Mastboom (2018) ¹²⁰	Diseases of the musculoskeletal system or connective tissue	Netherlands	0	0	337	Health-related quality of life
Corneloup (2020) ¹²¹	Diseases of the immune system	France	0	0	336	Health-related quality of life
Valassi (2022) ¹²²	Endocrine, nutritional or metabolic diseases	Multinational	0	0	320	Symptoms and diagnosis
Depping (2021) ¹²³	Groups of diseases	Germany	0	0	304	Expectations

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Shalhub (2020) ¹²⁴	Developmental anomalies	USA	0	0	300	Self-management
Pignolo (2020) ¹²⁵	Diseases of the musculoskeletal system or connective tissue	Multinational	0	0	299	Health-related quality of life
Wuyts (2018) ¹²⁶	Diseases of the respiratory system	Multinational	0	0	277	Health-related quality of life
Schuster (2022) ¹²⁷	Diseases of the nervous system	Multinational	0	0	275	Expectations
Hoyer (2022) ¹²⁸	Diseases of the respiratory system	Denmark	0	0	264	Health-related quality of life
Büttner (2022) ¹²⁹	Endocrine, nutritional or metabolic diseases	Germany	0	0	264	Health-related quality of life
Guilbert (2021) ¹³⁰	Groups of diseases	Spain	0	0	261	Care and support
Doser (2020) ¹³¹	Developmental anomalies	Denmark	0	0	244	Health-related quality of life
Schut (2022) ¹³²	Neoplasms	Multinational	0	0	235	Health-related quality of life
Francisco (2020) ¹³³	Endocrine, nutritional or metabolic diseases	Portugal	0	0	209	Quality of life
Magliano (2021) ¹³⁴	Endocrine, nutritional or metabolic diseases	Italy	0	0	200	Quality of life
Maqhuzu (2020) ¹³⁵	Diseases of the respiratory system	Germany	0	0	194	Health-related quality of life
Ragusa (2020) ¹³⁶	Developmental anomalies	Italy	0	0	193	Quality of life
Graziano (2022) ¹³⁷	Diseases of the respiratory system	Italy	0	0	192	Health-related quality of life
Dinur (2020) ¹³⁸	Endocrine, nutritional or metabolic diseases	Israel	0	0	192	Quality of life
Warby (2019) ¹⁴⁰	Neoplasms	Australia	0	0	184	Decision making
Nicoloro-SantaBarbara (2017) ¹³⁹	Neoplasms	USA	0	0	180	Quality of life
Bernthal (2021) ¹⁴⁰	Diseases of the musculoskeletal system or connective tissue	Multinational	0	0	166	Health-related quality of life
Anghelina (2022) ¹⁴¹	Neoplasms	USA	0	0	165	Health-related quality of life
Dimitrova (2021) ¹⁴²	Endocrine, nutritional or metabolic diseases	Switzerland	0	0	158	Health-related quality of life
Rihm (2022) ¹⁴³	Multiple rare diseases	Germany	0	0	149	Anxiety
Keller (2021) ¹⁴⁴	Endocrine, nutritional or metabolic diseases	Multinational	0	0	148	Health-related quality of life
Marques (2019) ¹⁴⁵	Developmental anomalies	Multinational	0	0	143	Care and support
Fjermestad (2018) ¹⁴⁶	Developmental anomalies	Norway	0	0	142	Health-related quality of life
Yuan (2020) ¹⁴⁷	Endocrine, nutritional or metabolic diseases	Netherlands	0	0	121	Health-related quality of life
Zöllner (2021) ¹⁴⁸	Developmental anomalies	Germany	0	0	121	Health-related quality of life
Witt (2019) ¹⁴⁹	Developmental anomalies	Germany	0	0	120	Health-related quality of life
Kuemmerle-Deschner (2020) ¹⁵⁰	Diseases of the immune system	Multinational	0	0	117	Health-related quality of life
Asperti (2022) ¹⁵¹	Diseases of the immune system	Italy	0	0	116	Anxiety
Weidema (2020) ^{152,153}	Neoplasms	Multinational	0	0	115	Care and support
Hanisch (2019) ¹⁵⁴	Developmental anomalies	Germany	0	0	110	Health-related quality of life
Friedlander (2019) ¹⁵⁵	Groups of diseases	France	0	0	110	Health-related quality of life
Junker (2021) ¹⁵⁶	Diseases of the nervous system	Multinational	0	0	109	Health-related quality of life
Darmaun (2021) ¹⁵⁷	Developmental anomalies	France	0	0	108	Health-related quality of life
Polistena (2021) ¹⁵⁸	Endocrine, nutritional or metabolic diseases	Italy	0	0	106	Health-related quality of life

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Liu (2018) ¹⁵⁹	Endocrine, nutritional or metabolic diseases	USA	0	0	106	Treatment and management
Wiegand-Grefe (2022) ¹⁶⁰	Multiple rare diseases	Germany	0	0	100	Health-related quality of life
Schmidt (2022) ¹⁶¹	Neoplasms	Germany	0	0	98	Health-related quality of life
van de Loo (2022) ¹⁶²	Groups of diseases	Netherlands	0	0	95	Health-related quality of life
Post (2021) ¹⁶³	Diseases of the nervous system	Multinational	0	0	95	Expectations
Chighizola (2021) ¹⁶⁴	Diseases of the immune system	Italy	0	0	92	Expectations
Bonnekoh (2022) ¹⁶⁵	Diseases of the skin	Germany	0	0	87	Health-related quality of life
Hyvönen (2020) ¹⁶⁶	Developmental anomalies	Finland	0	0	80	Health-related quality of life
Hanisch (2020) ¹⁶⁷	Developmental anomalies	Germany	0	0	79	Health-related quality of life
Lauby (2019) ¹⁶⁸	Diseases of the respiratory system	France	0	0	78	Health-related quality of life
Ramprasad (2021) ¹⁶⁹	Diseases of the nervous system	USA	0	0	77	Symptoms and side effects
Kodra (2020) ¹⁷⁰	Developmental anomalies	Italy	0	0	76	Health-related quality of life
Custers (2018) ¹⁷¹	Diseases of the nervous system	Netherlands	0	0	76	Quality of life
Lee (2021) ¹⁷²	Diseases of the immune system	Canada	0	0	72	Health-related quality of life
Saad (2021) ¹⁷³	Developmental anomalies	France	0	0	72	Health-related quality of life
Thouvenin (2021) ¹⁷⁴	Diseases of the visual system	France	0	0	72	Health-related quality of life
Rabenstein (2021) ¹⁷⁵	Diseases of the nervous system	Germany	0	0	71	Treatment and management
Mengel (2020) ¹⁷⁶	Endocrine, nutritional or metabolic diseases	Germany	0	0	69	Symptoms and diagnosis
Theodore-Oklota (2022) ¹⁷⁷	Developmental anomalies	Multinational	0	0	68	Quality of life
Reisner (2022) ¹⁷⁸	Diseases of the skin	USA	0	0	66	Health-related quality of life
Camarata (2021) ¹⁷⁹	Endocrine, nutritional or metabolic diseases	USA	0	0	62	Health-related quality of life
Camarata (2022) ¹⁸⁰	Endocrine, nutritional or metabolic diseases	Multi-national	0	0	62	Health-related quality of life
Hahn (2018) ¹⁸¹	Diseases of the immune system	Germany	0	0	61	Treatment and management
Underbjerg (2018) ¹⁸²	Endocrine, nutritional or metabolic diseases	Denmark	0	0	57	Health-related quality of life
De Sautu De Borbón (2021) ¹⁸³	Developmental anomalies	Spain	0	0	57	Health-related quality of life
Bahmer (2018) ¹⁸⁴	Diseases of the immune system	Germany	0	0	57	Treatment and management
Halimi (2018) ¹⁸⁵	Diseases of the circulatory system	France	0	0	55	Health-related quality of life
Biener (2021) ¹⁸⁶	Diseases of the immune system	Germany	0	0	54	Health-related quality of life
Hanisch (2018) ¹⁸⁷	Developmental anomalies	Germany	0	0	51	Symptoms and diagnosis
Xu (2021) ¹¹	Diseases of the immune system	Australia	0	0	50	Health-related quality of life
Yanes (2022) ¹⁸⁸	Endocrine, nutritional or metabolic diseases	Spain	0	0	50	Health-related quality of life
Crescimanno (2019) ¹⁸⁹	Diseases of the nervous system	Italy	0	0	48	Health-related quality of life
Defabianis (2022) ¹⁹⁰	Developmental anomalies	Italy	0	0	48	Health-related quality of life

Author (Year)	Disease category	Country	Interviews	Focus group	Surveys	Main focus
Murali (2022) ¹⁹¹	Endocrine, nutritional or metabolic diseases	USA	0	0	47	Health-related quality of life
Izquierdo-García (2020) ¹⁹²	Endocrine, nutritional or metabolic diseases	Spain	0	0	46	Care and support
Oelerich (2020) ¹⁹³	Developmental anomalies	Germany	0	0	46	Health-related quality of life
Hanisch (2020) ¹⁹⁴	Diseases of the digestive system	Germany	0	0	44	Health-related quality of life
Hanisch (2019) ¹⁹⁵	Endocrine, nutritional or metabolic diseases	Germany	0	0	43	Health-related quality of life
Maffi (2022) ¹⁹⁶	Diseases of the nervous system	Italy	0	0	42	Symptoms and side effects
Konradi (2021) ^{197,198}	Diseases of the musculoskeletal system or connective tissue	USA	0	0	39	Quality of life
Peltola (2021) ¹⁹⁹	Neoplasms	France	0	0	38	Health-related quality of life
Chiu (2022) ²⁰⁰	Multiple rare diseases	Hong Kong	0	0	36	Health-related quality of life
Gjørup (2021) ²⁰¹	Endocrine, nutritional or metabolic diseases/ Developmental anomalies	Denmark	0	0	35	Health-related quality of life
Herman (2022) ²⁰²	Endocrine, nutritional or metabolic diseases	Slovenia	0	0	34	Covid
Nguyen (2019) ²⁰³	Diseases of the circulatory system	USA	0	0	33	Health-related quality of life
Grimwood (2018) ²⁰⁴	Diseases of the digestive system	France	0	0	33	Health-related quality of life
Quijada-Fraile (2021) ²⁰⁵	Endocrine, nutritional or metabolic diseases	Spain	0	0	33	Symptoms and side effects
van de Loo (2022) ²⁰⁶	Endocrine, nutritional or metabolic diseases	Netherlands	0	0	33	Health-related quality of life
Carey (2021) ²⁰⁷	Developmental anomalies	USA	0	0	30	Symptoms and side effects
Kettenbach (2021) ²⁰⁸	Diseases of the immune system	Germany	0	0	30	Symptoms and side effects
Ashtari (2022) ²⁰⁹	Developmental anomalies	Multi-national	0	0	30	Online support
Holopainen (2019) ²¹⁰	Developmental anomalies	Finland	0	0	26	Symptoms and side effects
Stöberl (2019) ²¹¹	Developmental anomalies	Switzerland	0	0	24	Symptoms and side effects
Harmon (2019) ²¹²	Developmental anomalies	USA	0	0	22	Health-related quality of life
Niekamp (2020) ²¹³	Developmental anomalies	Germany	0	0	21	Health-related quality of life
Morrison (2019) ²¹⁴	Endocrine, nutritional or metabolic diseases	Multinational	0	0	13	Quality of life
Lechevalier (2022) ²¹⁵	Diseases of the skin	France	0	0	11	Care and support
Planellas (2021) ²¹⁶	Endocrine, nutritional or metabolic diseases	Spain	0	0	6	Symptoms and side effects

* Analysis of social media comments number of participants not given

Abbreviations and terminology

ASGS	The Australian Statistical Geography Standard from the Australian Bureau of Statistics, defines remoteness and urban/rural definitions in Australia
CCDR	Centre for Community-Driven Research
dF	Degrees of Freedom. The number of values in the final calculation of a statistic that are free to vary.
f	The F ratio is the ratio of two mean square values, used in an ANOVA comparison. A large F ratio means that the variation among group means is more than you'd expect to see by chance.
FOP	Fear of Progression. Tool to measure anxiety related to progression
IQR	Interquartile range. A measure of statistical dispersion, being equal to the difference between 75th and 25th percentiles, or between upper and lower quartiles.
p	Probability value. A small <i>p</i> -value (typically ≤ 0.05) indicates strong. A large <i>p</i> -value (> 0.05) indicates weak evidence.
PEEK	Patient Experience, Expectations and Knowledge
PIH	Partners in Health
SD	Standard deviation. A quantity expressing by how much the members of a group differ from the mean value for the group/
SEIFA	Socio-Economic Indexes for Areas (SEIFA) ranks areas in Australia according to relative socio-economic advantage and disadvantage. This is developed by the Australian Bureau of Statistics.
SF36	Short Form Health Survey 36
t	t-Statistic. Size of the difference relative to the variation in your sample data.
Tukey HSD	Tukey's honestly significant difference test. It is used in this study to find 9 significantly different means following an ANOVA test.
W	The W statistic is the test value from the Wilcoxon Rank sum test. The theoretical range of W is between 0 and (number in group one) x (number in group 2). When W=0, the two groups are exactly the same.
χ^2	Chi-squared. Kruskal-Wallis test statistic approximates a chi-square distribution. The Chi-square test is intended to test how likely it is that an observed distribution is due to chance.

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