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REVIEW

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European Reference Networks – a flagship activity of the EU in the field of rare and complex diseases: from 2017 to 2025

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Abstract

Background Although individual rare and complex diseases (RDs) affect small patient populations, together they impact an estimated 27–36 million people across the European Union. Addressing this major public health challenge has been a long-term priority for the European Union, leading to the establishment of the European Reference Networks (ERNs) in 2017.

Main body ERNs are cross-border networks connecting clinical expert centres to share knowledge, improve and harmonise diagnosis and care for patients with rare and complex diseases. Since their inception, 24 ERNs have united 1,606 expert centres across 375 hospitals in all EU Member States and Norway. Their activities span multidisciplinary clinical collaboration, patient-centred governance, education and training, and the development of clinical guidelines. Over 4900 extremely rare or difficult cases have been discussed among experts without requiring the patients to travel abroad when expertise was not available in their own countries. A key factor for this success is the cross-border IT platform - known as the Clinical Patient Management System 2.0 - provided by the European Commission for medical discussions, which enables experts to share patient data, including medical images and lab results, in a secure and protected environment that is fully compliant with all relevant security and data privacy requirements. ERNs have demonstrated resilience in crises such as the COVID-19 pandemic and the war in Ukraine, providing rapid, coordinated responses to sustain care for vulnerable patient groups. The first formal evaluation in 2023 confirmed that more than 95% of member centres met quality standards, underscoring the networks' maturity and effectiveness. Moving into the next phase, the Joint Action JARDIN (2024–2027) aims to integrate ERNs into national healthcare systems to ensure sustainability and equitable access to high-quality RD care.

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Conclusions ERNs exemplify European solidarity and innovation in healthcare, transforming how rare disease expertise is shared and applied across borders. Their continued integration into national systems will be pivotal to achieving a truly cohesive European Health Union that delivers improved outcomes for all patients with rare and complex diseases.

Keywords European Reference Networks, Rare diseases, Cross-border healthcare, European health union, Clinical patient management system, JARDIN, Patient-centred care, EU4Health

Whilst rare and complex diseases (RD) are uncommon, the number of citizens affected by them are not: in the EU alone, estimations are that between 27 and 36 million people live with a rare disease. Significant resources have therefore been deployed over time by the European Commission to address this key public health issue and support patients and families affected by a rare disease.

The European Commission has been active in addressing rare diseases for more than 30 years, first identified as a public health priority in the Commission in November 1993.

EU policy and action in the field of rare diseases is legally underpinned and guided by:

- The Commission Communication on 'Rare diseases: Europe's challenges'[1],
- The Council Recommendation on an action in the field of rare diseases[2],
- The Directive 2011/24/EU on patients' rights in cross-border healthcare[3].

One of the true European success stories that comes from this foundation are the 24 European Reference Networks (ERNs). These are cross-border networks that bring together centres of expertise and reference in different European hospitals to help patients. ERNs enable specialists in Europe to share expertise on cases of patients affected by rare, low-prevalence and complex diseases, providing advice on the most appropriate diagnosis and the best treatment available.

Together, these networks leverage the collective expertise for patient's care and are a prime example of European solidarity and innovation, allowing the expertise to travel, rather than patients.

The added value of the ERNs' daily work demonstrates the critical need for multidisciplinary care in rare and complex diseases. Within and across networks, ERNs facilitate collaboration across medical disciplines to ensure comprehensive patient-centred care. Joint sessions at scientific congresses are regularly organised to discuss advances in rare disease research and care. In parallel, ERN project management teams, funded by the EU, work collectively to disseminate information across defined disease areas, reinforcing collaboration, sharing resources, and enhancing training to improve patient care throughout Europe. All 24 ERNs are also working closely with disease-specific patient advocacy groups and associations to ensure that patients' perspectives remain at the centre of the activities.

Thanks to EU4Health programme[4] funding amounting to 77.4 million euro, the ERNs, at present, bring together 1.605 expert centres working in 375 hospitals located across the 27 Member States of the European Union and Norway. As such, ERNs have raised the level of collaboration and interaction to an unprecedented level, both in terms of quantity and quality.

In 2023, 24 European Reference Networks, including 836 members, completed their first evaluation[5]. Overall, the evaluation concluded that the ERN ecosystem is functioning well, providing highly specialist deliverables for rare disease patients such as consultations for diagnosis and therapies, the production of clinical guidelines and specialised trainings.

100% of the ERN networks and 88% of their members obtained satisfactory results, while only 4% of the members saw their membership terminated and 8% (72 Clinical Centres) were asked to submit an improvement plan. After the re-evaluation process in 2025, a total of 799 centres out of 836 evaluated clinical centres successfully passed the first periodic evaluation, which is about 95.6%. The membership of 37 (4.4%) clinical centres was terminated. Ultimately, the evaluation confirms the Networks have delivered on many areas of specialist work since their establishment. Since 2017, when the ERNs were officially established, the ERN system can be broadly divided into three phases.

[1] Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases - Europe's challenges.

[2] Council Recommendation of 8 June 2009 on an action in the field of rare diseases.

[3] Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare.

[4] EU4Health programme 2021–2027 – a vision for a healthier European Union - European Commission.

[5] Commission Implementing Decision 2014/287/EU of 10 March 2014 'setting out criteria for establishing and evaluating European Reference Networks and their Members and for facilitating the exchange of information and expertise on establishing and evaluating such Networks', as amended by the Commission Implementing Decision (EU) 2019/1269 of 26 July 2019, and in particular Article 12 and Article 14 thereof.

Phase one was a process of *learning-by-doing*, which – among others - involved the ERNs, the European Commission, the EU Member States, patient organisations, hospitals and clinicians. During this time, the overall ERN system began to take shape and develop, creating the innovative EU structure that we have today.

After this initial phase, the ERN system evolved into phase two: *consolidation*. The focus shifted towards strengthening inter-ERN coordination, ensuring homogeneity of core activities and integrating key performance indicators to help showcase the impact of the Networks' activities.

The system now stands at the beginning of phase three, which will address the primary challenge of achieving the full potential of the European Reference Networks: *integration of the ERNs into the national healthcare systems.* To this end, the Commission launched the Joint Action JARDIN to integrate the ERNs into national healthcare systems and help shape Union policies and programmes, and support Member States in the elaboration of national strategies. This should also facilitate the integration of the ERNs in the national healthcare systems to improve the care, diagnosis and treatment of rare disease patients and families. The Joint Action has a budget of 18.75 million Euros, covering a period of three years 2024–2027 and involves all EU Member States, Norway, and Ukraine as well as all key stakeholders involved in this field, such as patients' organisations and hospital managers.

To highlight the need for sustainability and integration of the ERNs' work, expertise and impact, this article provides an overview of the infrastructures the ERNs have been establishing and the different activities and initiatives they have been pursuing.

Healthcare and clinical patient management system

In 2017, the first year of the ERNs' existence, the centres participating in the ERNs indicated that they supported 161,379 patients with the diagnosis of a disease or condition within their medical scope. By 2024, this number increased to 416,234 patients, marking a growth of 160% over the course of 6 years of activities.

Alongside facilitating patient referrals, the Commission has financed the creation of the Clinical Patient Management System (CPMS). This tool is a secure and dedicated IT platform, that facilitates the work of the ERNs to provide cross-border clinical expertise by supporting cross-border medical discussions among healthcare professionals. In 2025, the Commission updated the tool to version 2.0 which enables a customised, more flexible, secure and user-friendly environment for healthcare professionals to share patient information, discuss cases and develop joint treatment plans. It also improves the quality and timeliness of patient care, supports medical imaging, improved data quality and data analysis whilst being fully compliant with the General Data Protection Regulation. The platform is also accessible via a dedicated portable application. As of February 2025, all ERNs are now using the new version of the IT platform.

Over the first seven years of their existence, the Networks' experts have discussed and provided multidisciplinary EU cross-border medical discussions on diagnosis, treatment and disease management for more than 4.900 extremely rare or difficult cases via the Clinical Patient Management System. This has allowed expertise, rather than people, to travel across borders for the delivery of high-quality care in respective care pathways [1–14]. CPMS uptake currently varies across ERNs. Structural clinical use-case-based implementation has been achieved by some ERNs with diagnosis, therapy decision and rare case constellation being key use cases. Tangible clinical decision benefits are reported. Key barriers include technical complexity, time/workflow burden, national data-protection requirements, and different training/helpdesk support.

The Clinical Patient Management System also allows early career experts to gain invaluable experience by participating in case discussions led by senior experts, learning how to prioritise investigations and treatments to achieve optimal patient outcomes.

With successful tools in place, the system for cross-border medical discussions among experts can now be leveraged to its full extent.

Patient centeredness

The ERNs put patient needs at the centre of their activities [15–66]. Patient involvement in ERNs takes place through the 24 European Patient Advocacy Groups (ePAGs), aligned to each ERN, with advocates formally embedded in ERN boards/working groups to ensure patient priorities inform governance, outputs and evaluation activities. Empirical work shows how this translates into practice: patient advocates participating in working groups on the development of guidelines, usage of large multi-country patient surveys to inform about education and quality improvement [56] as well as co-design and co-production of patient journeys[6]. However, implementation is variable across ERNs, with challenges including workload/time constraints for patient representatives, language, and uneven support structures for participation. Furthermore, there is a strong collaboration with EURORDIS – a non-profit alliance of over 1000 rare disease patient organisations from 74 countries – alongside a large number of patient organisations.

[6] https://www.ern-rnd.eu/disease-knowledge-hub/ern-rnd-patient-journeys/.

Patient advocacy organisations also act as educational resources. Patient advocates can learn more about disease diagnosis, treatment, and research methodologies. The presence of these organisations ensures that patients' perspectives are not only listened to, but integrated into the clinical, organisational and strategic decision-making processes of ERNs. Patient representatives actively contribute to the definition of diagnostic and therapeutic pathways, the validation of educational tools and the construction of models of cross-border care. Their contribution is even more relevant because it is anchored to the direct experience of life with the disease, and therefore capable of directing health choices towards a real centrality of the person.

For example, nearly 150 scientifically validated information leaflets and Patient Journeys were developed across the ERNs in collaboration with patient associations, providing an invaluable knowledge resource for patients and family doctors. The ePAG led co-production of Patient Journey leaflets, which are info-graphical overviews of rare disease patients' needs, and which are available in all EU languages, is a key achievement of ERNs. All such scientifically validated documentation can be freely downloaded from the ERNs' websites and used by physicians and caregivers.

Training and education

Rare and complex diseases present significant challenges in the education and training of healthcare professionals due to their low prevalence, limited evidence-based knowledge, and the rapid evolution of diagnostic tools, techniques, and treatment discoveries. Since their establishment in 2017, the 24 ERNs have worked on: (i) identifying and addressing training gaps and requirements; (ii) encouraging and facilitating the development of training and continuous education programs for early career experts and other healthcare providers within and beyond the Networks; (iii) organizing teaching and training activities, including exchanges; and (iv) developing and contributing to patient education strategies [67–88].

Leading clinicians, scientists, and healthcare providers within ERNs contributed their expertise to producing scientifically validated training and education resources for healthcare professionals. To disseminate knowledge, all ERNs developed open-access webinars presented by leading European experts. In the years 2020–2024, these activities have surged, resulting in more than 1,030 educational webinars, many incorporating multidisciplinary presentations. These webinars, now widely utilized in general hospital teaching sessions, exemplify state-of-the-art education for healthcare providers. In addition, more than 540 face-to-face educational activities, including workshops, seminars, and seasonal schools, were organized during this period. The European Commission also funded pilot exchange programs enabling short-term immersions in accredited clinical teams, fostering knowledge exchange, skills development, and cross-border collaboration. Over 350 individuals participated in these programs, tailored to the specificities of each ERN.

Structuring training activities in a sustainable fashion, ERNs have been establishing postgraduate training programs to educate the new generation of experts, thereby reflecting European training requirements that are adopted and approved by the European Union of Medical Specialties and respectively appraised examinations.

A robust network of trust and collaboration among clinical teams, underpinned by shared scientific practices, is now in place to provide cutting edge training and education activities across Europe.

ERNs increasingly link training "reach" metrics (participants, countries, materials) to effectiveness measures such as learning analytics and post-activity evaluations. Sustainability is supported by re-usable digital formats and will be facilitated by establishing digital databases and stronger integration into national training structures.

Clinical patient guidelines and other clinical decision support tools

In the years 2020–2024, the ERNs have also endorsed or developed more than 530 guidelines and recommendations, tailored to specific rare diseases to standardise care and enhance education across Member States. These guidelines significantly contribute to improving clinical practices [12, 89–337]. Additionally, ERNs provided evidence to policymakers to advocate for equitable access to diagnostic tools, such as genetic screening, and treatments [16, 145, 338–374].

ERNs in emergency situations

During their 7 years of existence, ERNs faced two major crises: the COVID-19 pandemic and Russia's war of aggression against Ukraine. In these extraordinary scenarios, the 24 Networks and their members concentrated their effort in setting up and participating in initiatives aimed at supporting both patients and their caregivers and healthcare professionals dealing with rare diseases [375–415]. Generally, ERNs promptly reacted by creating a COVID-19 emergency webpage in their websites and tailored their communication strategy to immediately become a hub for the rare disease community.

In the case of the Russia's war of aggression against Ukraine, the ERN coordinators spontaneously decided to initiate a concerted action to support patients from Ukraine: already in March 2022, they deployed an almost instant response via a devoted website [erncare4ua.com] to help Ukrainian patients contact the 24 coordinating ERN teams liaising with their members and relevant patient organisations for specialised medical support. In

addition to remote support, hundreds of cases have been referred to ERN medical teams which successfully organised multidisciplinary visits for affected Ukrainian refugees and provided support to healthcare professionals for the treatment of their patients located in Ukraine. Similarly to the COVID-19 emergency, all ERNs published a dedicated webpage on their websites focusing on assistance in their area of expertise and produced tailored educational products for healthcare professionals and patient advocates. A webinar series has been produced with simultaneous translation for Ukrainian patients and clinicians, including contributions from Ukrainian doctors.

This crisis-response experience highlights ERNs as a resilience asset. Key lessons are the importance of "digital-first" coordination, pre-defined escalation pathways, and better linkage between ERNs and national emergency/referral systems to reduce variability in access during shocks.

Looking forward, via the Joint Action for the Integration of ERNs into national healthcare systems (jardin-ern.eu), ERNs are now proposing to draft a suggested process for identifying crisis or emergency situations that may require a concrete and harmonised response for the benefit of the whole rare disease community.

Registries

Additional core activities, that lie within the healthcare scope of the ERNs, are the set-up of patient registries that contain interoperable patients' data seen by the ERNs. Registries allow patients' data to be harmonised via defined classification and ontologies (e.g., Orphanet for the disease classification or disease-specific ontologies) to enable the identification of patient cohorts for clinical research across different countries, identification of statistically significant cohorts for rare diseases and the generation of epidemiological information [416–462]. Furthermore, the registries function as a crucial platform linking care to research.

ERN registries apply GDPR-compliant data governance and include data access processes for secondary use. ERN registries align with the EU RD Platform (JRC)[7] building blocks, including the Set of Common Data Elements (EU-CDEs) and ERDRI tools (e.g., ERDRI.mdr metadata repository to support semantic interoperability). Links to broader European research infrastructures are being strengthened, for example with the ERN registry FAIRification [463] and interoperability support embedded in initiatives such as the European Joint Programming for RD [464] and the European Rare Disease Research Alliance (ERDERA).

ERNs also act as a key structure to allow the collection of large enough populations of patients with rare diseases through international European collaboration (between others SOLVE-RD [solve-rd.eu]; ERICA [erica-rd.eu]; ERDERA [erdera.org]; RealiseD [realised-ihi.eu]) to facilitate clinical research and trials.

In ERNs, cross-border *clinical* data exchange is primarily handled through the GDPR-aligned CPMS IT platform for medical discussions; FAIRification and (in some networks) federated querying are being developed mainly in the *registry/research* layer [430], but these are not yet systematically implemented across all ERNs, and federated learning remains an emerging rather than standardised ERN-wide approach.

Conclusion

The health impact of ERNs can be comparable to ensuring life-saving drug accessibility and foster sustainability, capacity building and empowerment on the national level. They also help to address critical healthcare issues such as availability, accessibility and quality of diagnostic practices including newborn screening.

Furthermore, ERN impact is increasingly approached through outcome-oriented measurement aligned to diagnostic pathways, clinical practice and cross-border access to expertise. A key enabler is the annual ERN continuous monitoring framework[8], which collects harmonised indicators across all ERNs, supporting longitudinal comparisons across networks and Member States[9]. At network level, concrete outcome signals are increasingly emerging linked to specific ERN activities such as CPMS based use cases, ERN registry data collection and analysis [416], as well as implementation of healthcare quality cycles [465]. Also, cross-ERN registry harmonisation efforts are beginning to operationalise pathway outcomes such as time to diagnosis, utilising interoperable data elements of all ERN registries. Today, the ERNs represent well-structured and highly interconnected networks that have built strong collaborative relationships with the respective European professional societies. Although still in the consolidation phase, ERNs are already having a significant impact on how care is being provided to patients with rare diseases in Europe. The true potential of ERNs, however, will only be reached when the ERNs are more efficiently linked to the national health systems. That is the aim of the three-year Joint Action JARDIN.

Ultimately, ERNs epitomise the concrete added value and successful results that can come from strong European collaboration and networking in health. ERNs lie

[7] https://eu-rd-platform.jrc.ec.europa.eu.

[8] ERN Continuous Monitoring and Quality Improvement System - Public Health.

[9] https://health.ec.europa.eu/publications/european-reference-networks-monitoring-report-2025_en.

Addendum 1 Websites of all 24 European Reference Networks

Acronym	Name	Website
Endo-ERN	European Reference Network on rare endocrine conditions	https://endo-ern.eu/
ERKNet	European Reference Network on rare kidney diseases	https://www.erknet.org/
ERN BOND	European Reference Network on rare bone disorders	https://ernbond.eu/
ERN CRANIO	European Reference Network on rare craniofacial anomalies and ear, nose and throat (ENT) disorders	https://www.ern-cranio.eu/
ERN EpiCARE	European Reference Network on rare and complex epilepsies	https://epi-care.eu/
ERN EURACAN	European Reference Network on rare adult solid cancers	https://www.euracan.eu/
ERN eUROGEN	European Reference Network on rare uro-recto-genital diseases and complex conditions	https://eurogen-ern.eu/
ERN EURO-NMD	European Reference Network on rare neuromuscular diseases	https://ern-euro-nmd.eu/
ERN GENTURIS	European Reference Network on rare genetic tumour risk syndromes	https://www.genturis.eu/
ERN GUARD-Heart	European Reference Network on uncommon and rare diseases of the heart	https://guardheart.ern-net.eu/
ERN PaedCan	European Reference Network on paediatric cancer (haemato-oncology)	https://paedcan.ern-net.eu/
ERN RARE-LIVER	European Reference Network on rare hepatological diseases	https://rare-liver.eu/
ERN ReCONNET	European Reference Network on rare connective tissue and musculoskeletal diseases	https://reconnet.ern-net.eu/
ERN RITA	European Reference Network on rare immunodeficiency, autoinflammatory, autoimmune, and paediatric rheumatic diseases	https://ern-rita.org/
ERN TRANSPLANT-CHILD	European Reference Network on transplantation in children	https://transplantchild.eu/
ERN-EuroBloodNet	European Reference Network on rare haematological diseases	https://eurobloodnet.eu/
ERN-EYE	European Reference Network on rare eye diseases	https://www.ern-eye.eu/
ERN-ITHACA	European Reference Network on rare malformation syndromes, intellectual and other neuro-developmental disorders	https://ern-ithaca.eu/
ERN-LUNG	European Reference Network on rare respiratory diseases	https://ern-lung.eu/
ERN-RND	European Reference Network on rare neurological diseases	https://www.ern-rnd.eu/
ERN-Skin	European Reference Network on rare, complex, and undiagnosed skin disorders	https://ern-skin.eu/
ERNICA	European Reference Network on rare inherited and congenital (digestive and gastrointestinal) anomalies	https://www.ern-ernica.eu/
MetabERN	European Reference Network on hereditary metabolic disorders	https://metab.ern-net.eu/
VASCERN	European Reference Network on rare multisystemic vascular diseases	https://vascern.eu/

at the heart of a strong European Health Union, which aims to improve the health of all citizens, no matter where they live, no matter their disease or complex condition. The accumulated experience and the cross-border dimension of the ERNs place them as a flagship initiative in the European Health Union.

Abbreviations

COVID-19	Coronavirus Disease
CPMS	Clinical Patient Management System
ePAG	European Patient Advocacy Group
ERN	European Reference Network
EU	European Union
JARDIN	Joint Action for the Integration of ERNs into national healthcare systems
RD	Rare and Complex Diseases

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None of the authors declare any competing interests.

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References

1. Schoenmakers DH, Asbreuk M, Martin T, Datema M, Beerepoot S, Inbar-Feigenberg M, et al. Key lessons from the first international treatment eligibility committee: the case of metachromatic leukodystrophy. Eur J Paediatr Neurol. 2025;57:72–81.
2. Joneborg U, Bergamini A, Wallin E, Mangili G, Solheim O, Marquina G, et al. European multidisciplinary tumor boards support cross-border networking and increase treatment options for patients with rare gynecological tumors. Int J Gynecol Cancer. 2023;33(10):1621–6.
3. Fortunato F, Bianchi F, Ricci G, Torri F, Gualandi F, Neri M, et al. Digital health and Clinical Patient Management System (CPMS) platform utility for data sharing of neuromuscular patients: the Italian EURO-NMD experience. Orphanet J Rare Dis. 2023;18(1):196.
4. El-Hassar L, Amara A, Sanson B, Lacatus O, Amir Belhouchet A, Kroneman M, et al. Telemedicine in Neuromuscular Diseases During Covid-19 Pandemic: ERN-NMD European Survey. J Neuromuscul Dis. 2023;10(2):173–84.
5. White EK, Wagner IV, van Beuzekom C, Iotova V, Ahmed SF, Hiort O, et al. A critical evaluation of the EU-virtual consultation platform (CPMS) within the European Reference Network on Rare Endocrine Conditions. Endocr Connect. 2022;11(11).
6. Talarico R, Marinello D, Cannizzo S, Palla I, Ticciati S, Gaglioti A, et al. An Opportunity to Harmonise the Approach to Patients' Care Pathways for Rare and Complex Diseases: RarERN Path™. Front Health Serv. 2022;2:935014.
7. Talarico R, Aguilera S, Alexander T, Amoura Z, Andersen J, Arnaud L, et al. The added value of a European Reference Network on rare and complex connective tissue and musculoskeletal diseases: insights after the first 5 years of the ERN ReCONNET. Clin Exp Rheumatol. 2022;40(Suppl 134):3–11.
8. Tumiene B, Graessner H, Mathijssen IM, Pereira AM, Schaefer F, Scarpa M, et al. European Reference Networks: challenges and opportunities. J Community Genet. 2021;12(2):217–29.
9. Talarico R, Cannizzo S, Lorenzoni V, Marinello D, Palla I, Pirri S, et al. Correction to: RarERN Path: a methodology towards the optimisation of patients' care pathways in rare and complex diseases developed within the European Reference Networks. Orphanet J Rare Dis. 2021;16(1):146.
10. Paglialonga A, Gaetano R, Robert L, Hurard M, Botella LM, Barr N, et al. eHealth for patients with rare diseases: the eHealth Working Group of the European Reference Network on Rare Multisystemic Vascular Diseases (VASCERN). Orphanet J Rare Dis. 2021;16(1):164.
11. Mönig I, Steenvoorden D, de Graaf JP, Ahmed SF, Taruscio D, Beun JG, et al. CPMS-improving patient care in Europe via virtual case discussions. Endocrine. 2021;71(3):549–54.
12. Walther D, Steinmann O, Wagner TOF, Storf H. Implementation of EXABO - An Expert Advisory Board for the European Reference Network for Rare Respiratory Diseases. Stud Health Technol Inf. 2020;270:986–90.
13. Smith M, Alexander E, Marcinkute R, Dan D, Rawson M, Banka S, et al. Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet J Rare Dis. 2020;15(1):103.
14. Walther D, Steinmann O, Schaefer J, Wagner TOF, Storf H. Conception of an Expert Advisory Board for the European Reference Network for Rare Respiratory Diseases. Stud Health Technol Inf. 2018;247:236–40.
15. Spivack OKC, Klein Haneveld MJ, Louisse S, Slater G, Hernando I. Supporting the continuous development and use of a patient partnership framework in European rare disease networks (ERNs): a scoping review of frameworks in the scientific literature. J Community Genet. 2025;16(1):15–28.
16. Zucchi D, Marinello D, Tani C, Fulvio G, Aguilera S, Benachi A, et al. Pregnancy-related issues in rare and low-prevalence diseases: results of ERN transversal working group on pregnancy and family planning survey. Orphanet J Rare Dis. 2025;20(1):112.
17. Tschamper MK, Wahl AK, Jakobsen R, Larsen MH. Meeting Parents' Health Literacy Needs-Designing and Feasibility Testing of a Three-Step Intervention in the Childhood Epilepsy Context. J Clin Nurs. 2025;34(11):4479–98.
18. Padilla CS, Bergerot CD, Dijke K, Roets E, Boková G, Innerhofer V, et al. Health-Related Quality of Life (HRQoL) assessments in research on patients with adult rare solid cancers: a state-of-the-art review. Cancers (Basel). 2025;17(3).
19. Faasse M, Groot-Sluijsmans BC, Coopmans AG, van de Bovenkamp HM. FACE ME'-The Impact and Value of an Arts-Based Project About the Patient-Parent-Clinician Relationship in European Reference Network CRANIO. J Craniofac Surg. 2025;36(8):3004–12.
20. Engels M, Urbanczyk K, Hölzenspies J, Röhl C, Geverink N, Hoogerbrugge N. The European Reference Network on Genetic Tumour Risk Syndromes (ERN

GENTURIS): benefits for patients, families, and health care providers. Fam Cancer. 2025;24(2):33.
21. Domínguez-Carral J, Reinhard C, Soliani L, Cif L, Ortigoza-Escobar JD. Exploring the Impact of Dyskinetic Crises in GNAO1-Related Disorders: A Survey for Parents and Caregivers. Mov Disord Clin Pract. 2025;12(10):1615–21.
22. Wiethoff I, Sikking M, Evers S, Gabrio A, Henkens M, Michels M, et al. Quality of life and societal costs in patients with dilated cardiomyopathy. Eur Heart J Qual Care Clin Outcomes. 2024;10(4):334–44.
23. van Pottelberghe S, Kupper N, Scheirlynck E, Amin AS, Wilde AAM, Hofman N, et al. Are disease-specific patient-reported outcomes measures (PROMs) used in cardiogenetics? A systematic review. Eur J Hum Genet. 2024;32(6):607–18.
24. Soto Jansson J, Bjurulf B, Dellenmark Blom M, Hallböök T, Reilly C. Diagnosis, epilepsy treatment and supports for neurodevelopment in children with Dravet Syndrome: Caregiver reported experiences and needs. Epilepsy Behav. 2024;151:109603.
25. Soto Jansson J, Bjurulf B, Dellenmark Blom M, Hallböök T, Reilly C. Caregiver perceptions of the impact of Dravet syndrome on the family, current supports and hopes and fears for the future: A qualitative study. Epilepsy Behav. 2024;156:109790.
26. Scognamiglio D, Boarini M, la Forgia MC, Grippa E, Forni S, Sergi A, et al. Defining priorities in the transition from paediatric to adult healthcare for rare bone disease patients: a dialogic approach. Eur J Med Genet. 2024;67:104891.
27. Leroy BP, Daly A, Héon E, Sahel JA, Dollfus H. Therapies for Inherited Retinal Dystrophies: What is Enough? Drug Discov Today. 2024;29(9):104095.
28. Kassabian B, Levy AM, Gardella E, Aledo-Serrano A, Ananth AL, Brea-Fernández AJ, et al. Developmental epileptic encephalopathy in DLG4-related synaptopathy. Epilepsia. 2024;65(4):1029–45.
29. Chemaly N, Kuchenbuch M, Teng T, Marie E, D'Onofrio G, Lo Barco T, et al. A European pilot study in Dravet Syndrome to delineate what really matters for the patients and families. Epilepsia Open. 2024;9(1):388–96.
30. Zamanipoor Najafabadi AH, van der Meulen M, Priego Zurita AL, Faisal Ahmed S, van Furth WR, Charmandari E, et al. Starting point for benchmarking outcomes and reporting of pituitary adenoma surgery within the European Reference Network on Rare Endocrine Conditions (Endo-ERN): results from a meta-analysis and survey study. Endocr Connect. 2023;12(1).
31. Violani C, Grano C, Fernandes M, Prato AP, Feitz WFJ, Wijnen R, et al. The Transition of Care for Patients with Anorectal Malformations and Hirschsprung Disease: A European Survey. Eur J Pediatr Surg. 2023;33(3):191–7.
32. van Roessel I, de Graaf JP, Biermasz NR, Charmandari E, van Santen HM. Acquired hypothalamic dysfunction in childhood: 'what do patients need?' - an Endo-ERN survey. Endocr Connect. 2023;12(10).
33. Roganovic J, Bien E, Ferrari A, Vassal G, Trama A, Casali PG, et al. Solutions for optimal care and research for children and adolescents with extremely rare cancers developed within the Joint Action for Rare Cancers (JARC). EJC Pediatr Oncol. 2023;2.
34. Pellegrini M, Chakravorty S, Del Mar Manu Pereira M, Gulbis B, Gilmour-Hamilton C, Hayes S, et al. Sickle cell disease: embedding patient participation into an international conference can transform the role of lived experience. Orphanet J Rare Dis. 2023;18(1):341.
35. Nott E, Behl KE, Brambilla I, Green TE, Lucente M, Vavassori R, et al. Rare. The importance of research, analysis, reporting and education in 'solving' the genetic epilepsies: A perspective from the European patient advocacy group for EpiCARE. Eur J Med Genet. 2023;66(3):104680.
36. de Beaufort CMC, Aminoff D, de Blaauw I, Crétolle C, Dingemann J, Durkin N, et al. Transitional Care for Patients with Congenital Colorectal Diseases: An EUPSA Network Office, ERNICA, and eUROGEN Joint Venture. J Pediatr Surg. 2023;58(12):2319–26.
37. Beun JG, Burman P, Kämpe O, Husebye ES, Hahner S, Kristensen J, et al. Doctors, teach your adrenal insufficiency patients well: provide them with a European Emergency Card! Endocr Connect. 2023;12(1).
38. Wu CI, Schwartz PJ, Ackerman MJ, Wilde AAM. COVID-19 vaccination in patients with long QT syndrome. Heart Rhythm O2. 2022;3(6):706–9.
39. Webb SM, Kristensen J, Nordenström A, Vitali D, Amodru V, Wiehe LK, et al. Patient journey experiences may contribute to improve healthcare for patients with rare endocrine diseases. Endocr Connect. 2022;11(12).
40. Russo FM, Debeer A, De Coppi P, Devriendt K, Crombag N, Hubble T, et al. What should we tell parents? Congenital diaphragmatic hernia. Prenat Diagn. 2022;42(3):398–407.
41. Platero L, Garcia-Sanchez P, Sainz T, Calvo C, Iglesias I, Esperon F, et al. Pets for pediatric transplant recipients: To have or not to have. Front Vet Sci. 2022;9:974665.
42. Peltonen S, Jannic A, Wolkenstein P. Treatment of cutaneous neurofibromas with carbon dioxide laser: Technique and patient experience. Eur J Med Genet. 2022;65(1):104386.
43. Marinello D, Galetti I, Dan D, Andersson AS, Aguilera S, Louisse S, et al. Patient engagement in healthcare: a preliminary set of measures to evaluate patient engagement in the European Reference Networks. Rare Disease Orphan Drugs J. 2022;1(1).
44. Iotova V, Bertherat J, Mastorakos G, Hiort O, Pereira AM, ENDO-ERN ON, Rare endocrine conditions: Endo-ERN in its fifth year: a pinch of care, science, curiosity and new horizons. Endocr Connect. 2022;11(3).
45. Höhn S, Dozières-Puyravel B, Auvin S. Expectations of patient associations from a French Center for Rare Epilepsies. Epilepsy Behav. 2022;136:108927.
46. Davidse K, van Staa A, Geilvoet W, van Eck JP, Pellikaan K, Baan J, et al. We mind your step: understanding and preventing drop-out in the transfer from paediatric to adult tertiary endocrine healthcare. Endocr Connect. 2022;11(5).
47. Bjurulf B, Reilly C, Hallböök T. Caregiver reported seizure precipitants and measures to prevent seizures in children with Dravet syndrome. Seizure. 2022;103:3–10.
48. Benson M, Albanese A, Bhatia KP, Cavillon P, Cuffe L, König K, et al. Development of a patient journey map for people living with cervical dystonia. Orphanet J Rare Dis. 2022;17(1):130.
49. Webb SM, Kristensen J, Vitali D, van Klink S, van Beuzekom C, Santos A, et al. EndoERN patient survey on their perception of health care experience and of unmet needs for rare endocrine diseases. Endocrine. 2021;71(3):569–77.
50. Stepien KM, Kieć-Wilk B, Lampe C, Tangeraas T, Cefalo G, Belmatoug N, et al. Challenges in Transition From Childhood to Adulthood Care in Rare Metabolic Diseases: Results From the First Multi-Center European Survey. Front Med (Lausanne). 2021;8:652358.
51. Smit M, Albanese A, Benson M, Edwards MJ, Graessner H, Hutchinson M, et al. Dystonia Management: What to Expect From the Future? The Perspectives of Patients and Clinicians Within DystoniaNet Europe. Front Neurol. 2021;12:646841.
52. Roussel E, Peeters E, Vanthoor J, Bozzini G, Muneer A, Ayres B, et al. Predictors of local recurrence and its impact on survival after glansectomy for penile cancer: time to challenge the dogma? BJU Int. 2021;127(5):606–13.
53. Pascau MJ, Pruneda L, de Barbieri I, Correia M, López B, Guijarro E, et al. Social resources for transplanted children and families in European union hospitals of ERN TransplantChild. Child (Basel). 2021;8(9).
54. Kristensen J, Brösamle M, van den Berg B. Patient's view on better care. Endocrine. 2021;71(3):706–7.
55. Jürgensen M, Rapp M, Döhnert U, Frielitz FS, Ahmed F, Cools M, et al. Assessing the health-related management of people with differences of sex development. Endocrine. 2021;71(3):675–80.
56. Iotova V, Schalin-Jäntti C, Bruegmann P, Broesamle M, De Graaf J, Bratina N, et al. Access to patient oriented information-a baseline Endo-ERN survey among patients with rare endocrine disorders. Endocrine. 2021;71(3):542–8.
57. Drewitz KP, Grey J, Brügmann P, Pichl J, Sammarco M, Aarts M, et al. Patients' perception on the quality of care for multiple endocrine neoplasia disorders in Europe: an online survey from a patient support group. Endocrine. 2021;71(3):634–40.
58. de Graaf JP, de Vries F, Dirkson A, Hiort O, Pereira AM, Korbonits M, et al. Patients with rare endocrine conditions have corresponding views on unmet needs in clinical research. Endocrine. 2021;71(3):561–8.
59. Conde Blanco E, Manzanares I, Centeno M, Khawaja M, Betrán O, Donaire A, et al. Epilepsy and lockdown: A survey of patients normally attending a Spanish centre. Acta Neurol Scand. 2021;143(2):206–9.
60. Claessen K, Andela CD, Biermasz NR, Pereira AM. Clinical Unmet Needs in the Treatment of Adrenal Crisis: Importance of the Patient's Perspective. Front Endocrinol (Lausanne). 2021;12:701365.
61. Wu CI, Postema PG, Arbelo E, Behr ER, Bezzina CR, Napolitano C, et al. SARS-CoV-2, COVID-19, and inherited arrhythmia syndromes. Heart Rhythm. 2020;17(9):1456–62.
62. Trivisano M, Specchio N, Pietrafusa N, Calabrese C, Ferretti A, Ricci R, et al. Impact of COVID-19 pandemic on pediatric patients with epilepsy - The caregiver perspective. Epilepsy Behav. 2020;113:107527.
63. de Vries F, Bruin M, Cersosimo A, van Beuzekom CN, Ahmed SF, Peeters RP, et al. An overview of clinical activities in Endo-ERN: the need for alignment of future network criteria. Eur J Endocrinol. 2020;183(2):141–8.
64. Bolz-Johnson M, Meek J, Hoogerbrugge N. Patient Journeys: improving care by patient involvement. Eur J Hum Genet. 2020;28(2):141–3.

65. Vos JR, Giepmans L, Röhl C, Geverink N, Hoogerbrugge N. Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. Fam Cancer. 2019;18(2):281–4.
66. Kreuzer M, Drube J, Prüfe J, Schaefer F, Pape L. Current management of transition of young people affected by rare renal conditions in the ERKNet. Eur J Hum Genet. 2019;27(12):1783–90.
67. Weber MT, Noll R, Marchl A, Facchinello C, Grünewaldt A, Hügel C, et al. MedBot vs RealDoc: efficacy of large language modeling in physician-patient communication for rare diseases. J Am Med Inf Assoc. 2025;32(5):775–83.
68. van Eerde AM, Teixeira A, Galletti F, Maternik M, Capone V, Westland R, et al. Risks and benefits of ChatGPT in informing patients and families with rare kidney diseases: an explorative assessment by the European Rare Kidney Disease Reference Network (ERKNet). Pediatr Nephrol. 2025;40(9):2899–905.
69. Frauscher B, Mansilla D, Abdallah C, Astner-Rohracher A, Beniczky S, Brazdil M, et al. Learn how to interpret and use intracranial EEG findings. Epileptic Disord. 2024;26(1):1–59.
70. Berger A, Grimm KL, Noll R, Wagner TO. Pareto-principle in rare disease education: assessing the representation of common rare diseases in medical education and coding systems. Orphanet J Rare Dis. 2024;19(1):340.
71. Meeting abstracts from the 12th European Conference on Rare Diseases and Orphan Products. Orphanet Journal of Rare Diseases. 2024;19(S1).
72. Iotova V, Schalin-Jäntti C, Van Beuzekom C, Bruegmann P, Broesamle M, Hiort O, et al. An overview of the outreach of the 2019–2021 Endo-ERN knowledge generation webinars. Endocr Connect. 2023;12(9).
73. Dargaud Y, Fontana P, Fenaux P, Hermans C. Specialist training in thrombosis and haemostasis across Europe: From aspirations to actions. Haemophilia. 2023;29(5):1369–70.
74. Centen LM, Pinter D, van Egmond ME, Graessner H, Kovacs N, Koy A, et al. Dystonia management across Europe within ERN-RND: current state and future challenges. J Neurol. 2023;270(2):797–809.
75. Meeting abstracts from the 11th edition of the European conference on Rare Diseases & Orphan Products (ECRD) 2022. Orphanet J Rare Dis. 2023;18(Suppl 1):118.
76. Tumiene B, Peters H, Melegh B, Peterlin B, Utkus A, Fatkulina N, et al. Rare disease education in Europe and beyond: time to act. Orphanet J Rare Dis. 2022;17(1):441.
77. Nascimento FA, Jing J, Strowd R, Sheikh IS, Weber D, Gavvala JR, et al. Competency-based EEG education: a list of must-know EEG findings for adult and child neurology residents. Epileptic Disord. 2022;24(5):979–82.
78. Lesca G, Baumgartner T, Monin P, De Dominicis A, Kunz WS, Specchio N. Genetic causes of rare and common epilepsies: What should the epileptologist know? Eur J Med Genet. 2022;65(9):104570.
79. Farhat MM, Cornet A, Frank C, Galetti I, Grunert J, Guimarães V, et al. Exploring patient education unmet needs for rare and complex connective tissue and musculoskeletal diseases: A survey of health care providers' and patients' expectations in Europe. Chronic Illn. 2022;18(2):370–80.
80. Dourado E, Melo AT, Pacini G, Pankow A, Klemm P, Cruz-Machado AR. Rheumatology residency in a European Reference Network on Rare and Complex Connective Tissue and Musculoskeletal Diseases centre: opportunities and challenges. Clin Exp Rheumatol. 2022;40(Suppl 134):63–5.
81. Brunelle Praschberger A, Post AEM, Hermanns S, Graessner H. Establishing and boosting communication in the European Reference Network for Rare Neurological Diseases (ERN-RND): the impact of offering free educational webinars. Orphanet J Rare Dis. 2022;17(1):89.
82. Blümcke I, Biesel E, Bedenlier S, Händel M, Wilmshurst J, Mehndiratta MM, et al. A structured, blended learning program towards proficiency in epileptology: the launch of the ILAE Academy Level 2 Program. Epileptic Disord. 2022;24(5):737–50.
83. Iotova V, Schalin-Jäntti C, Bruegmann P, Broesamle M, Bratina N, Tillmann V, et al. Educational and knowledge gaps within the European reference network on rare endocrine conditions. Endocr Connect. 2021;10(1):37–44.
84. Beniczky S, Blümcke I, Rampp S, Shisler P, Biesel E, Wiebe S. e-learning comes of age: Web-based education provided by the International League Against Epilepsy. Epileptic Disord. 2020;22(3):237–44.
85. 10th European Conference on Rare Diseases & Orphan Products (ECRD 2020). Orphanet J Rare Dis. 2020;15(Suppl 1):310.
86. Powell P, Kreuter M, Wijsenbeek-Lourens M. Where are the gaps in education in the field of rare lung disease? Perspectives from the ERN-LUNG educational programme survey. Breathe (Sheff). 2019;15(2):102–3.
87. Blümcke I, Arzimanoglou A, Beniczky S, Wiebe S. Roadmap for a competency-based educational curriculum in epileptology: report of the Epilepsy Education Task Force of the International League Against Epilepsy. Epileptic Disord. 2019;21(2):129–40.
88. Murray S, Labbé S, Kothare S, Málaga I, Kluger G, Ogden P, et al. Identifying the educational needs of physicians in pediatric epilepsy in order to improve care: results from a needs assessment in Germany, Spain, and the United States. Epileptic Disord. 2018;20(4):239–56.
89. Vanegas MI, Marcé-Grau A, Cazurro-Gutiérrez A, Hamami F, Timmers ER, Weißbach A, et al. Approach to Myoclonus Dystonia Syndrome: A European Reference Network Survey. Mov Disord Clin Pract. 2025;12(7):957–68.
90. van Roey VL, van der Mink AB, Mathijssen IMJ, Akota I, de Blacam C, Breugem CC, et al. Between unity and disparity: current treatment protocols for common orofacial clefts in European expert centres. Int J Oral Maxillofac Surg. 2025;54(6):519–28.
91. Torra R, Lipska-Zietkiewicz B, Acke F, Antignac C, Becker JU, Cornec-Le Gall E, et al. Diagnosis, management and treatment of the Alport syndrome – 2024 guideline on behalf of ERKNet, ERA and ESPN. Nephrol Dial Transpl. 2025;40(6):1091–106.
92. Schoenmakers DH, Beerepoot S, Adang LA, Asbreuk MABC, Bergner CG, Bley AE, et al. Gene therapy in advanced metachromatic leukodystrophy: tempering expectations. Protein Cell. 2025;16(1):12–5.
93. Rifino N, Hervè D, Acerbi F, Kuroda S, Lanzino G, Vajkoczy P, et al. Diagnosis and management of adult Moyamoya angiopathy: An overview of guideline recommendations and identification of future research directions. Int J Stroke. 2025;20(5):512–23.
94. Ribrag V, Bron D, Rymkiewicz G, Hoelzer D, Jørgensen J, de Armas-Castellano A, et al. Diagnosis and treatment of Burkitt lymphoma in adults: clinical practice guidelines from ERN-EuroBloodNet. Lancet Haematol. 2025;12(2):e138–50.
95. Resteghini C, Baujat B, Bossi P, Franchi A, de Gabory L, Halamkova J, et al. Sinonasal malignancy: ESMO-EURACAN Clinical Practice Guideline for diagnosis, treatment and follow-up. ESMO Open. 2025;10(2):104121.
96. Michel K, Ruiz-Ramos A, Nou-Fontanet L, Martín-Gomez C, Carmona Hidalgo B, Isabel-Gomez R, et al. Respiratory and other organ manifestations in NKX2-1-related disorders: a systematic review. Front Med (Lausanne). 2025;12:1507513.
97. Mancuso M, Lopriore P, Semmler L, Kornblum C. 280th ENMC International Workshop: The ERN EURO-NMD mitochondrial diseases working group; diagnostic criteria and outcome measures in primary mitochondrial myopathies. Hoofddorp, the Netherlands, 22–24 November 2024. Neuromuscul Disord. 2025;50:105340.
98. Levtchenko E, Ariceta G, Arguedas Flores O, Bichet DG, Bockenhauer D, Emma F, et al. International expert consensus statement on the diagnosis and management of congenital nephrogenic diabetes insipidus (arginine vasopressin resistance). Nat Rev Nephrol. 2025;21(2):83–96.
99. Kruse MTA, Olde Dubbelink BAS, Kroneman M, de Groot I, Schlüter S, de Visser M, et al. Awareness of bone strength in patients with neuromuscular disorders: ERN EURO-NMD clinician survey and European patient survey. J Neurol Sci. 2025;472:123420.
100. Kremer MEB, Gorter RR, Tidman J, Gosemann JH, de Blaauw I. Development of European Consensus Guidelines and Evolution of Innovative Surgical Strategies for the Treatment of Anorectal Malformations. Eur J Pediatr Surg. 2025;35(2):77–8.
101. Krämer S, Hillebrecht AL, Bekes K, Bücher K, Clark V, Haririan H, et al. Oral health care pathways for patients with epidermolysis bullosa: A position statement from the European reference network for rare skin diseases. J Eur Acad Dermatol Venereol. 2025;39(6):1080–90.
102. Jurcut R, Barriales-Villa R, Biagini E, Garcia-Pavia P, Olivotto I, Protonotarios A, et al. Key priorities for the implementation of the 2023 ESC Guidelines for the management of cardiomyopathies in low-resource settings. Eur Heart J Qual Care Clin Outcomes. 2025;11(7):910–8.
103. Granja-Dominguez A, Martin-Gomez C, Ortigoza-Escobar JD, Rodriguez-Lopez R, Gonzalez-Bermudez L, Dantone S, et al. A proposal to involve people living with rare and complex conditions in the development of clinical practice guidelines. Patient Educ Couns. 2025;135:108708.
104. Engel B, Assis DN, Bhat M, Clusmann J, Drenth JP, Gerussi A, et al. Quo vadis autoimmune hepatitis? - Summary of the 5(th) international autoimmune hepatitis group research workshop 2024. JHEP Rep. 2025;7(2):101265.
105. El Hachem M, Caldaro T, Lara-Corrales I, Pope E, Jones R, Bageta ML, et al. Management of oesophageal strictures in inherited epidermolysis bullosa: a clinical practice guideline. Br J Dermatol. 2025;193(3):394–404.
106. Della-Torre E, Talarico R, Ballarin J, Bozzalla-Cassione E, Cardamone C, Cigolini C, et al. Identification of red flags for IgG4-related disease: an international

European Reference Network for Rare Connective Tissue Diseases framework. Lancet Rheumatol. 2025;7(1):e64–71.
107. de Armas-Castellano A, Infante-Ventura D, Del Pino-Sedeño T, Hernández YG, Quiros R, León-Salas B, et al. Prognostic Factors for Survival in Adults With Burkitt Lymphoma: A Systematic Review. Cancer Med. 2025;14(3):e70513.
108. Cannizzo S, Quoidbach V, Trieste L, Benson M, Federico A, Filla A, et al. The organizational dimension in rare and complex diseases care management: an application of RarERN Path(©) methodology in ataxias, dystonia and phenylketonuria. BMC Health Serv Res. 2025;25(1):799.
109. Bökenkamp A, Ariceta G, Böckenhauer D, Devuyst O, Emma F, van Bennekom D, et al. Dent disease: clinical practice recommendations. Nephrol Dial Transpl. 2025;40(5):852–64.
110. Aubert O, Irvine WFE, Aminoff D, de Blaauw I, Cascio S, Cretolle C, et al. European Reference Network eUROGEN Guidelines on the Management of Anorectal Malformations, Part II: Treatment. Eur J Pediatr Surg. 2025;35(2):112–9.
111. Aubert O, Irvine WFE, Aminoff D, de Blaauw I, Cascio S, Cretolle C, et al. ERN eUROGEN Guidelines on the Management of Anorectal Malformations Part I: Diagnostics. Eur J Pediatr Surg. 2025;35(2):104–11.
112. Aubert O, Irvine W, Aminoff D, de Blaauw I, Cascio S, Cretolle C, et al. ERN eUROGEN Guidelines on the Management of Anorectal Malformations, Part IV: Organization of Care and Communication between Providers. Eur J Pediatr Surg. 2025;35(2):128–34.
113. Arnaud L, Ruiz-Irastorza G, Aranow C, Bernatsky S, Dall'Era M, Adelowo O, et al. ERN ReCONNET-SLICC-SLEuro expert consensus on the therapeutic management of rare systemic lupus erythematosus manifestations. Lancet Rheumatol. 2025;7(7):e505–18.
114. EASL-ERN Clinical Practice Guidelines on Wilson's disease. J Hepatol. 2025.
115. Zaffaroni G, Mannucci A, Koskenvuo L, de Lacy B, Maffioli A, Bisseling T, et al. Updated European guidelines for clinical management of familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP), gastric adenocarcinoma, proximal polyposis of the stomach (GAPPS) and other rare adenomatous polyposis syndromes: a joint EHTG-ESCP revision. Br J Surg. 2024;111(5).
116. Wittebrood C, Boban M, Cagnin A, Capellari S, De Winter FL, Djamshidian A, et al. Pharmacotherapy for behavioural manifestations in frontotemporal dementia: An expert consensus from the European Reference Network for Rare Neurological Diseases (ERN-RND). Eur J Neurol. 2024;31(12):e16446.
117. Wege H, Campani C, de Kleine R, Meyer T, Nault JC, Pawlik TM, et al. Rare primary liver cancers: An EASL position paper. J Hepatol. 2024.
118. Weber MT, Schaaf J, Storf H, Wagner TOF, Berger A, Noll R. Editing Physicians' Responses Using GPT-4 for Academic Research. Stud Health Technol Inf. 2024;313:101–6.
119. Verdonschot JAJ, Hellebrekers D, van Empel VPM, Heijligers M, de Munnik S, Coonen E, et al. Clinical Guideline for Preimplantation Genetic Testing in Inherited Cardiac Diseases. Circ Genom Precis Med. 2024;17(2):e004416.
120. Van Damme P, Al-Chalabi A, Andersen PM, Chiò A, Couratier P, De Carvalho M, et al. European Academy of Neurology (EAN) guideline on the management of amyotrophic lateral sclerosis in collaboration with European Reference Network for Neuromuscular Diseases (ERN EURO-NMD). Eur J Neurol. 2024;31(6):e16264.
121. Valente KD, Reilly C, Carvalho RM, Smith ML, Mula M, Wirrell EC, et al. Consensus-based recommendations for the diagnosis and treatment of anxiety and depression in children and adolescents with epilepsy: A report from the Psychiatric Pediatric Issues Task Force of the International League Against Epilepsy. Epilepsia. 2024;65(11):3155–85.
122. Tetart F, Walsh S, Milpied B, Gaspar K, Vorobyev A, Tiplica GS, et al. Acute generalized exanthematous pustulosis: European expert consensus for diagnosis and management. J Eur Acad Dermatol Venereol. 2024;38(11):2073–81.
123. Spier I, Yin X, Richardson M, Pineda M, Laner A, Ritter D, et al. Gene-specific ACMG/AMP classification criteria for germline APC variants: Recommendations from the ClinGen InSiGHT Hereditary Colorectal Cancer/Polyposis Variant Curation Expert Panel. Genet Med. 2024;26(2):100992.
124. Shibbani K, Abdulkarim A, Budts W, Roos-Hesselink J, Müller J, Shafer K, et al. Participation in Competitive Sports by Patients With Congenital Heart Disease: AHA/ACC and EAPC/ESC/AEPC Guidelines Comparison. J Am Coll Cardiol. 2024;83(7):772–82.
125. Servais A, Zacchia M, Dehoux L, Shroff R, Brassier A, Taurisano R, et al. Clinical Practice Recommendations on Kidney Management in Methylmalonic Acidemia: an Expert Consensus Statement From ERKNet and MetabERN. Kidney Int Rep. 2024;9(12):3362–74.
126. Schoenmakers DH, Mochel F, Adang LA, Boelens JJ, Calbi V, Eklund EA, et al. Inventory of current practices regarding hematopoietic stem cell transplantation in metachromatic leukodystrophy in Europe and neighboring countries. Orphanet J Rare Dis. 2024;19(1):46.
127. Revencu N, Eijkelenboom A, Bracquemart C, Alhopuro P, Armstrong J, Baselga E, et al. Assessment of gene-disease associations and recommendations for genetic testing for somatic variants in vascular anomalies by VASCERN-VASCA. Orphanet J Rare Dis. 2024;19(1):213.
128. Reijman MD, Kusters DM, Groothoff JW, Arbeiter K, Dann EJ, de Boer LM, et al. Clinical practice recommendations on lipoprotein apheresis for children with homozygous familial hypercholesterolaemia: An expert consensus statement from ERKNet and ESPN. Atherosclerosis. 2024;392:117525.
129. Ray-Coquard I, Casali PG, Croce S, Fennessy FM, Fischerova D, Jones R, et al. ESGO/EURACAN/GCIG guidelines for the management of patients with uterine sarcomas. Int J Gynecol Cancer. 2024;34(10):1499–521.
130. Pina-Garza JE, Chez M, Cloyd J, Hirsch LJ, Kälviäinen R, Klein P, et al. Outpatient management of prolonged seizures and seizure clusters to prevent progression to a higher-level emergency: Consensus recommendations of an expert working group. Epileptic Disord. 2024;26(4):484–97.
131. Perucca E, French JA, Aljandeel G, Balestrini S, Braga P, Burneo JG, et al. Which terms should be used to describe medications used in the treatment of seizure disorders? An ILAE position paper. Epilepsia. 2024;65(3):533–41.
132. Patel S, Maney K, Morris L, Papadopoulou MT, Prange L, Boggs A, et al. Real life retrospective study of cannabidiol therapy in alternating hemiplegia of childhood. Eur J Paediatr Neurol. 2024;49:55–9.
133. Parenti G, Fecarotta S, Alagia M, Attaianese F, Verde A, Tarallo A, et al. The European reference network for metabolic diseases (MetabERN) clinical pathway recommendations for Pompe disease (acid maltase deficiency, glycogen storage disease type II). Orphanet J Rare Dis. 2024;19(1):408.
134. Nou-Fontanet L, Nguyen QTR, Bachoud-Levi AC, Reinhard C, Ortigoza-Escobar JD. Insights from European Reference Network for rare neurological disorders study surveys on diagnosis, treatment, and management of NKX2-1-related disorders. Eur J Paediatr Neurol. 2024;51:110–7.
135. Neff MC, Schaaf J, Noll R, Holtz S, Schütze D, Köhler SM, et al. Initial User-Centred Design of an AI-Based Clinical Decision Support System for Primary Care. Stud Health Technol Inf. 2024;310:1051–5.
136. Nanetti L, Kearney M, Boesch S, Stovickova L, Ortigoza-Escobar JD, Macaya A, et al. Child-to-adult transition: a survey of current practices within the European Reference Network for Rare Neurological Diseases (ERN-RND). Neurol Sci. 2024;45(3):1007–16.
137. Muneer A, Bandini M, Compérat E, De Meerleer G, Fizazi K, Gietema J, et al. Penile cancer: ESMO-EURACAN Clinical Practice Guideline for diagnosis, treatment and follow-up. ESMO Open. 2024;9(7):103481.
138. Mulatero P, Scholl UI, Fardella CE, Charmandari E, Januszewicz A, Reincke M, et al. Familial hyperaldosteronism: an European Reference Network on Rare Endocrine Conditions clinical practice guideline. Eur J Endocrinol. 2024;190(4):G1–14.
139. Mekahli D, Müller RU, Marlais M, Wlodkowski T, Haeberle S, de Argumedo ML, et al. Clinical practice recommendations for kidney involvement in tuberous sclerosis complex: a consensus statement by the ERKNet Working Group for Autosomal Dominant Structural Kidney Disorders and the ERA Genes & Kidney Working Group. Nat Rev Nephrol. 2024;20(6):402–20.
140. McLin VA, Franchi-Abella S, Brütsch T, Bahadori A, Casotti V, de Ville de Goyet J, et al. Expert management of congenital portosystemic shunts and their complications. JHEP Rep. 2024;6(1):100933.
141. McDevitt T, Durkie M, Arnold N, Burghel GJ, Butler S, Claes KBM, et al. EMQN best practice guidelines for genetic testing in hereditary breast and ovarian cancer. Eur J Hum Genet. 2024;32(5):479–88.
142. Mazzolai L, Teixido-Tura G, Lanzi S, Boc V, Bossone E, Brodmann M, et al. 2024 ESC Guidelines for the management of peripheral arterial and aortic diseases. Eur Heart J. 2024;45(36):3538–700.
143. Maver A, Lohmann K, Borovečki F, Wolstenholme N, Taylor RL, Spielmann M, et al. Quality assurance for next-generation sequencing diagnostics of rare neurological diseases in the European Reference Network. Eur J Hum Genet. 2024;32(8):1014–21.
144. Mancuso M, Papadopoulou MT, Ng YS, Ardissone A, Bellusci M, Bertini E, et al. Management of seizures in patients with primary mitochondrial diseases: consensus statement from the InterERNs Mitochondrial Working Group. Eur J Neurol. 2024;31(7):e16275.
145. Mancuso M, Lopriore P, Lamperti C, Klopstock T, Rahman S, Licchetta L, et al. Current management of primary mitochondrial disorders in EU countries: the European Reference Networks survey. J Neurol. 2024;271(2):835–40.

146. Lugowska I, Becker JC, Ascierto PA, Veness M, Blom A, Lebbe C, et al. Merkel-cell carcinoma: ESMO-EURACAN Clinical Practice Guideline for diagnosis, treatment and follow-up. ESMO Open. 2024;9(5):102977.
147. Liu APY, Li BK, Vasiljevic A, Dewan MC, Tamrazi B, Ertl-Wagner B, et al. SNO-EANO-EURACAN consensus on management of pineal parenchymal tumors. Neuro Oncol. 2024;26(12):2159–73.
148. Lenarczyk R, Zeppenfeld K, Tfelt-Hansen J, Heinzel FR, Deneke T, Ene E, et al. Management of patients with an electrical storm or clustered ventricular arrhythmias: a clinical consensus statement of the European Heart Rhythm Association of the ESC-endorsed by the Asia-Pacific Heart Rhythm Society, Heart Rhythm Society, and Latin-American Heart Rhythm Society. Europace. 2024;26(4).
149. Lammers RJM, Tsachouridis G, Andersson MK, Dormeus S, Ekerhult TO, Frankiewicz M, et al. What should be next in lifelong posterior hypospadias: Conclusions from the 2023 ERN eUROGEN and EJP-RD networking meeting. Neurourol Urodyn. 2024;43(5):1097–103.
150. Israni M, Alderson E, Mahlaoui N, Obici L, Rossi-Semerano L, Lachmann H, et al. Recommendations for Transitioning Young People with Primary Immunodeficiency Disorders and Autoinflammatory Diseases to Adult Care. J Clin Immunol. 2024;45(1):57.
151. Hannema SE, Busiah K, Butler G, Claahsen-van de Grinten HL, Cools M, Gawlik-Starzyk AM, et al. Endocrine management of transgender and gender-diverse adolescents: expert opinion of the ESPE working group on gender incongruence and the Endo-ERN main thematic group on sexual development and maturation. Horm Res Paediatr. 2024:1–27.
152. Gravholt CH, Andersen NH, Christin-Maitre S, Davis SM, Duijnhouwer A, Gawlik A, et al. Clinical practice guidelines for the care of girls and women with Turner syndrome. Eur J Endocrinol. 2024;190(6):G53–151.
153. Granström AL, Irvine W, Hoel AT, Tabbers M, Kyrklund K, Fascetti-Leon F, et al. Ernica Clinical Consensus Statements on Total Colonic and Intestinal Aganglionosis. J Pediatr Surg. 2024;59(10):161565.
154. Geilswijk M, Genuardi M, Woodward ER, Nightingale K, Huber J, Madsen MG, et al. ERN GENTURIS clinical practice guidelines for the diagnosis, surveillance and management of people with Birt-Hogg-Dubé syndrome. Eur J Hum Genet. 2024;32(12):1542–50.
155. Fautrel B, Mitrovic S, De Matteis A, Bindoli S, Antón J, Belot A, et al. EULAR/PReS recommendations for the diagnosis and management of Still's disease, comprising systemic juvenile idiopathic arthritis and adult-onset Still's disease. Ann Rheum Dis. 2024;83(12):1614–27.
156. Drackley A, Somerville C, Arnaud P, Baudhuin LM, Hanna N, Kluge ML, et al. Interpretation and classification of FBN1 variants associated with Marfan syndrome: consensus recommendations from the Clinical Genome Resource's FBN1 variant curation expert panel. Genome Med. 2024;16(1):154.
157. Domínguez Carral J, Reinhard C, Ebrahimi-Fakhari D, Dorison N, Galosi S, Garone G, et al. Dyskinetic crisis in GNAO1-related disorders: clinical perspectives and management strategies. Front Neurol. 2024;15:1403815.
158. Dollfus H, Lilien MR, Maffei P, Verloes A, Muller J, Bacci GM, et al. Bardet-Biedl syndrome improved diagnosis criteria and management: Inter European Reference Networks consensus statement and recommendations. Eur J Hum Genet. 2024;32(11):1347–60.
159. Cuperus IE, Mathijssen IMJ, van Veelen MC, Bouzariouh A, Stubelius I, Kölby L, et al. A European Multicenter Outcome Study of Perioperative Airway Management Policies following Midface Surgery in Syndromic Craniosynostosis. Plast Reconstr Surg. 2024;154(6):1281–92.
160. Carmona-Hidalgo B, Herrera-Ramos E, Rodríguez-López R, Nou-Fontanet L, Blasco-Amaro JCM. Systematic review of thyroid function in NKX2-1-related disorders: Treatment and follow-up. PLoS ONE. 2024;19(10):e0309064.
161. Burgos CM, Irvine W, Vivanti A, Conner P, Machtejeviene E, Peters N, et al. European reference network for rare inherited congenital anomalies (ERNICA) evidence based guideline on the management of gastroschisis. Orphanet J Rare Dis. 2024;19(1):60.
162. Brüggen MC, Walsh S, Ameri MM, Anasiewicz N, Maverakis E, French LE, et al. Management of Adult Patients With Drug Reaction With Eosinophilia and Systemic Symptoms: A Delphi-Based International Consensus. JAMA Dermatol. 2024;160(1):37–44.
163. Blay JY, Casali P, Ray-Coquard I, Seckl MJ, Gietema J, de Herder WW, et al. Management of patients with rare adult solid cancers: objectives and evaluation of European reference networks (ERN) EURACAN. Lancet Reg Health Eur. 2024;39:100861.
164. Amato ME, Darling A, Stovickova L, Attard S, Eggink H, Engelen M, et al. Improving paediatric movement disorders care: Insights on rating scales utilization and clinical practice. Eur J Paediatr Neurol. 2024;52:10–9.
165. Al-Samkari H, Shehata N, Lang-Robertson K, Bianchi P, Glenthøj A, Sheth S, et al. Diagnosis and management of pyruvate kinase deficiency: international expert guidelines. Lancet Haematol. 2024;11(3):e228–39.
166. Vogel A, Bridgewater J, Edeline J, Kelley RK, Klümpen HJ, Malka D, et al. Biliary tract cancer: ESMO Clinical Practice Guideline for diagnosis, treatment and follow-up. Ann Oncol. 2023;34(2):127–40.
167. Van't Westeinde A, Karlsson L, Messina V, Wallensteen L, Brösamle M, Dal Maso G, et al. An update on the long-term outcomes of prenatal dexamethasone treatment in congenital adrenal hyperplasia. Endocr Connect. 2023;12(4).
168. Vallortigara J, Greenfield J, Hunt B, Hoffman D, Reinhard C, Graessner H, et al. Patient pathways for rare diseases in Europe: ataxia as an example. Orphanet J Rare Dis. 2023;18(1):328.
169. Terman SW, Slinger G, Rheaume CE, Haque AS, Smith SN, van Griethuysen R, et al. Antiseizure Medication Withdrawal Practice Patterns: A Survey Among Members of the American Academy of Neurology and EpiCARE. Neurol Clin Pract. 2023;13(1):e200109.
170. Téllez L, Payancé A, Tjwa E, Del Cerro MJ, Idorn L, Ovroutski S, et al. EASL-ERN position paper on liver involvement in patients with Fontan-type circulation. J Hepatol. 2023;79(5):1270–301.
171. Specchio N, Nabbout R, Aronica E, Auvin S, Benvenuto A, de Palma L, et al. Updated clinical recommendations for the management of tuberous sclerosis complex associated epilepsy. Eur J Paediatr Neurol. 2023;47:25–34.
172. Snijders R, Assis DN, Oo YH, Sebode M, Taubert R, Willemse J, et al. Research gaps and opportunities in autoimmune hepatitis-Results of the international autoimmune hepatitis group research workshop 2022. Liver Int. 2023;43(7):1375–84.
173. Sessa C, Balmaña J, Bober SL, Cardoso MJ, Colombo N, Curigliano G, et al. Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline. Ann Oncol. 2023;34(1):33–47.
174. Poitou C, Holland A, Höybye C, de Graaff LCG, Bottius S, Otterlei B, et al. The transition from pediatric to adult care in individuals with Prader-Willi syndrome. Endocr Connect. 2023;12(1).
175. Pellerino A, Verdijk RM, Nichelli L, Andratschke NH, Idbaih A, Goldbrunner R. Diagnosis and treatment of peripheral and cranial nerve tumors with expert recommendations: an EUropean Network for RAre CANcers (EURACAN) Initiative. Cancers (Basel). 2023;15(7).
176. Nijhuis WH, Verhoef M, Sakkers RJB. Life span care for patients with skeletal dysplasia: A roadmap. Eur J Med Genet. 2023;66(11):104851.
177. Nabbout R, Zanello G, Baker D, Black L, Brambilla I, Buske OJ, et al. Towards the international interoperability of clinical research networks for rare diseases: recommendations from the IRDiRC Task Force. Orphanet J Rare Dis. 2023;18(1):109.
178. Morris S, Vallortigara J, Greenfield J, Hunt B, Hoffman D, Reinhard C, et al. Impact of specialist ataxia centres on health service resource utilisation and costs across Europe: cross-sectional survey. Orphanet J Rare Dis. 2023;18(1):382.
179. Martín-Gómez C, Ortigoza-Escobar JD, Nou-Fontanet L, Molina-Linde JM, Bachoud-Lévi AC, Léger J, et al. Exploring the values, preferences, and information needs of patients with NKX2-1-related disorders: A qualitative study protocol. PLoS ONE. 2023;18(2):e0281573.
180. Lemelle L, Flaadt T, Fresneau B, Moya-Plana A, Timmermann B, Roganovic J, et al. NUT Carcinoma in Children and Adolescents: The Expert European Standard Clinical Practice Harmonized Recommendations. J Pediatr Hematol Oncol. 2023;45(4):165–73.
181. Lalloo F, Kulkarni A, Chau C, Nielsen M, Sheaff M, Steele J, et al. Clinical practice guidelines for the diagnosis and surveillance of BAP1 tumour predisposition syndrome. Eur J Hum Genet. 2023;31(11):1261–9.
182. Krishnan U, Dumont MW, Slater H, Gold BD, Seguy D, Bouin M, et al. The International Network on Oesophageal Atresia (INoEA) consensus guidelines on the transition of patients with oesophageal atresia-tracheoesophageal fistula. Nat Rev Gastroenterol Hepatol. 2023;20(11):735–55.
183. Krasemann T, Butera G, Thomson J, Chessa M, Bokenkamp R. AEPC recommendations for training in interventional catheterisation for CHD. Cardiol Young. 2023;33(6):854–61.
184. Groothoff JW, Metry E, Deesker L, Garrelfs S, Acquaviva C, Almardini R, et al. Clinical practice recommendations for primary hyperoxaluria: an expert consensus statement from ERKNet and OxalEurope. Nat Rev Nephrol. 2023;19(3):194–211.
185. Förster A, Davenport C, Duployez N, Erlacher M, Ferster A, Fitzgibbon J, et al. European standard clinical practice - Key issues for the medical care of individuals with familial leukemia. Eur J Med Genet. 2023;66(4):104727.

186. Fassnacht M, Assie G, Baudin E, Eisenhofer G, Fouchardiere C, Haak HR, et al. Corrigendum to Adrenocortical carcinomas and malignant phaeochromocytomas: ESMO-EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up: [Annals of Oncology volume 31 (2020) 1476–1490]. Ann Oncol. 2023;34(7):631.
187. Evangelista A, Sitges M, Jondeau G, Nijveldt R, Pepi M, Cuellar H, et al. Multimodality imaging in thoracic aortic diseases: a clinical consensus statement from the European Association of Cardiovascular Imaging and the European Society of Cardiology working group on aorta and peripheral vascular diseases. Eur Heart J Cardiovasc Imaging. 2023;24(5):e65–85.
188. De Giorgis V, Tagliabue A, Bisulli F, Brambilla I, Camerini A, Cusmai R, et al. Ketogenic dietary therapies in epilepsy: recommendations of the Italian League against Epilepsy Dietary Therapy Study Group. Front Neurol. 2023;14:1215618.
189. Damstra RJ, Vignes S, Mansour S. Consensus recommendations on lymphedema in Phelan-McDermid syndrome. Eur J Med Genet. 2023;66(6):104767.
190. Carton C, Evans DG, Blanco I, Friedrich RE, Ferner RE, Farschtschi S, et al. ERN GENTURIS tumour surveillance guidelines for individuals with neurofibromatosis type 1. EClinicalMedicine. 2023;56:101818.
191. Brignole M, Cecchi F, Anastasakis A, Crotti L, Deharo JC, Elliott PM, et al. Syncope in hypertrophic cardiomyopathy (part II): An expert consensus statement on the diagnosis and management. Int J Cardiol. 2023;370:330–7.
192. Bossi P, Chan AT, Even C, Machiels JP. ESMO-EURACAN Clinical Practice Guideline update for nasopharyngeal carcinoma: adjuvant therapy and first-line treatment of recurrent/metastatic disease. Ann Oncol. 2023;34(3):247–50.
193. Bersano A, Khan N, Fuentes B, Acerbi F, Canavero I, Tournier-Lasserve E, et al. European Stroke Organisation (ESO) Guidelines on Moyamoya angiopathy Endorsed by Vascular European Reference Network (VASCERN). Eur Stroke J. 2023;8(1):55–84.
194. Arbelo E, Protonotarios A, Gimeno JR, Arbustini E, Barriales-Villa R, Basso C, et al. [2023 ESC Guidelines for the management of cardiomyopathies]. G Ital Cardiol (Rome). 2023;24(11 Suppl 1):e1–127.
195. Andrade RJ, Aithal GP, de Boer YS, Liberal R, Gerbes A, Regev A, et al. Nomenclature, diagnosis and management of drug-induced autoimmune-like hepatitis (DI-ALH): An expert opinion meeting report. J Hepatol. 2023;79(3):853–66.
196. Aleksovska K, Bassetti CLA, Berger T, Carvalho V, Costa J, Deuschl G, et al. Prioritization process for European Academy of Neurology clinical practice guidelines. Eur J Neurol. 2023;30(2):305–20.
197. Zeppenfeld K, Tfelt-Hansen J, de Riva M, Winkel BG, Behr ER, Blom NA, et al. 2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death. Eur Heart J. 2022;43(40):3997–4126.
198. Wilde AAM, Semsarian C, Márquez MF, Shamloo AS, Ackerman MJ, Ashley EA, et al. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Europace. 2022;24(8):1307–67.
199. Verkouteren BJA, Cosgun B, Reinders M, Kessler P, Vermeulen RJ, Klaassens M, et al. A guideline for the clinical management of basal cell naevus syndrome (Gorlin-Goltz syndrome). Br J Dermatol. 2022;186(2):215–26.
200. van Herpen C, Vander Poorten V, Skalova A, Terhaard C, Maroldi R, van Engen A, et al. Salivary gland cancer: ESMO-European Reference Network on Rare Adult Solid Cancers (EURACAN) Clinical Practice Guideline for diagnosis, treatment and follow-up. ESMO Open. 2022;7(6):100602.
201. van Haalen FM, Kaya M, Pelsma ICM, Dekkers OM, Biermasz NR, Cannegieter SC, et al. Current clinical practice for thromboprophylaxis management in patients with Cushing's syndrome across reference centers of the European Reference Network on Rare Endocrine Conditions (Endo-ERN). Orphanet J Rare Dis. 2022;17(1):178.
202. van den Bersselaar LR, Heytens L, Silva HCA, Reimann J, Tasca G, Díaz-Cambronero Ó, et al. European Neuromuscular Centre consensus statement on anaesthesia in patients with neuromuscular disorders. Eur J Neurol. 2022;29(12):3486–507.
203. Specchio N, Wirrell EC, Scheffer IE, Nabbout R, Riney K, Samia P, et al. International League Against Epilepsy classification and definition of epilepsy syndromes with onset in childhood: Position paper by the ILAE Task Force on Nosology and Definitions. Epilepsia. 2022;63(6):1398–442.
204. Specchio N, Pietrafusa N, Perucca E, Cross JH. New paradigms for the treatment of pediatric monogenic epilepsies: Progressing toward precision medicine. Epilepsy Behav. 2022;131:107961. Pt B).
205. Souche E, Beltran S, Brosens E, Belmont JW, Fossum M, Riess O, et al. Recommendations for whole genome sequencing in diagnostics for rare diseases. Eur J Hum Genet. 2022;30(9):1017–21.
206. Rudà R, Capper D, Waldman AD, Pallud J, Minniti G, Kaley TJ, et al. EANO - EURACAN - SNO Guidelines on circumscribed astrocytic gliomas, glioneuronal, and neuronal tumors. Neuro Oncol. 2022;24(12):2015–34.
207. Plotkin SR, Messiaen L, Legius E, Pancza P, Avery RA, Blakeley JO, et al. Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation. Genet Med. 2022;24(9):1967–77.
208. Petzold A, Fraser CL, Abegg M, Alroughani R, Alshowaeir D, Alvarenga R, et al. Diagnosis and classification of optic neuritis. Lancet Neurol. 2022;21(12):1120–34.
209. Nordenström A, Ahmed SF, van den Akker E, Blair J, Bonomi M, Brachet C, et al. Pubertal induction and transition to adult sex hormone replacement in patients with congenital pituitary or gonadal reproductive hormone deficiency: an Endo-ERN clinical practice guideline. Eur J Endocrinol. 2022;186(6):G9–49.
210. Müller RU, Messchendorp AL, Birn H, Capasso G, Cornec-Le Gall E, Devuyst O, et al. An update on the use of tolvaptan for autosomal dominant polycystic kidney disease: consensus statement on behalf of the ERA Working Group on Inherited Kidney Disorders, the European Rare Kidney Disease Reference Network and Polycystic Kidney Disease International. Nephrol Dial Transpl. 2022;37(5):825–39.
211. Kohl S, Avni FE, Boor P, Capone V, Clapp WL, De Palma D, et al. Definition, diagnosis and clinical management of non-obstructive kidney dysplasia: a consensus statement by the ERKNet Working Group on Kidney Malformations. Nephrol Dial Transpl. 2022;37(12):2351–62.
212. Knoers N, Antignac C, Bergmann C, Dahan K, Giglio S, Heidet L, et al. Genetic testing in the diagnosis of chronic kidney disease: recommendations for clinical practice. Nephrol Dial Transpl. 2022;37(2):239–54.
213. Hirsch E, French J, Scheffer IE, Bogacz A, Alsaadi T, Sperling MR, et al. ILAE definition of the Idiopathic Generalized Epilepsy Syndromes: Position statement by the ILAE Task Force on Nosology and Definitions. Epilepsia. 2022;63(6):1475–99.
214. Hadady L, Klivényi P, Perucca E, Rampp S, Fabó D, Bereczki C, et al. Web-based decision support system for patient-tailored selection of antiseizure medication in adolescents and adults: An external validation study. Eur J Neurol. 2022;29(2):382–9.
215. Frappaz D, Dhall G, Murray MJ, Goldman S, Faure Conter C, Allen J, et al. EANO, SNO and Euracan consensus review on the current management and future development of intracranial germ cell tumors in adolescents and young adults. Neuro Oncol. 2022;24(4):516–27.
216. Evans DG, Mostaccioli S, Pang D, Fadzil OCM, Pittara M, Champollion N, et al. ERN GENTURIS clinical practice guidelines for the diagnosis, treatment, management and surveillance of people with schwannomatosis. Eur J Hum Genet. 2022;30(7):812–7.
217. Castelletti S, Gray B, Basso C, Behr ER, Crotti L, Elliott PM, et al. Indications and utility of cardiac genetic testing in athletes. Eur J Prev Cardiol. 2022;29(12):1582–91.
218. Casali PG, Blay JY, Abecassis N, Bajpai J, Bauer S, Biagini R, et al. Gastrointestinal stromal tumours: ESMO-EURACAN-GENTURIS Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol. 2022;33(1):20–33.
219. Capone V, Persico N, Berrettini A, Decramer S, De Marco EA, De Palma D, et al. Definition, diagnosis and management of fetal lower urinary tract obstruction: consensus of the ERKNet CAKUT-Obstructive Uropathy Work Group. Nat Rev Urol. 2022;19(5):295–303.
220. Aleksovska K, Kobulashvili T, Costa J, Zimmermann G, Ritchie K, Reinhard C, et al. European Academy of Neurology guidance for developing and reporting clinical practice guidelines on rare neurological diseases. Eur J Neurol. 2022;29(6):1571–86.
221. Weber LT, Tönshoff B, Grenda R, Bouts A, Topaloglu R, Gülhan B, et al. Clinical practice recommendations for recurrence of focal and segmental glomerulosclerosis/steroid-resistant nephrotic syndrome. Pediatr Transpl. 2021;25(3):e13955.
222. Wagner A, Aretz S, Auranen A, Bruno MJ, Cavestro GM, Crosbie EJ, et al. The Management of Peutz-Jeghers Syndrome: European Hereditary Tumour Group (EHTG) Guideline. J Clin Med. 2021;10(3).
223. Virgone C, Roganovic J, Vorwerk P, Redlich A, Schneider DT, Janic D, et al. Adrenocortical tumours in children and adolescents: The EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29025.

224. van Trotsenburg P, Stoupa A, Léger J, Rohrer T, Peters C, Fugazzola L, et al. Congenital Hypothyroidism: A 2020–2021 Consensus Guidelines Update-An ENDO-European Reference Network Initiative Endorsed by the European Society for Pediatric Endocrinology and the European Society for Endocrinology. Thyroid. 2021;31(3):387–419.
225. Tumiene B, Graessner H. Rare disease care pathways in the EU: from odysseys and labyrinths towards highways. J Community Genet. 2021;12(2):231–9.
226. Trepiccione F, Walsh SB, Ariceta G, Boyer O, Emma F, Camilla R, et al. Distal renal tubular acidosis: ERKNet/ESPN clinical practice points. Nephrol Dial Transpl. 2021;36(9):1585–96.
227. Surun A, Schneider DT, Ferrari A, Stachowicz-Stencel T, Rascon J, Synakiewicz A, et al. Salivary gland carcinoma in children and adolescents: The EXPeRT/PARTNER diagnosis and treatment recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29058.
228. Strauss SJ, Frezza AM, Abecassis N, Bajpai J, Bauer S, Biagini R, et al. Bone sarcomas: ESMO-EURACAN-GENTURIS-ERN PaedCan Clinical Practice Guideline for diagnosis, treatment and follow-up. Ann Oncol. 2021;32(12):1520–36.
229. Stiles MK, Wilde AAM, Abrams DJ, Ackerman MJ, Albert CM, Behr ER, et al. 2020 APHRS/HRS expert consensus statement on the investigation of decedents with sudden unexplained death and patients with sudden cardiac arrest, and of their families. J Arrhythm. 2021;37(3):481–534.
230. Stachowicz-Stencel T, Synakiewicz A, Cornet M, Ferrari A, Garassino M, Masip JR, et al. Thymoma and thymic carcinoma in children and adolescents: The EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29042.
231. Sestini S, Paneghetti L, Lampe C, Betti G, Bond S, Bellettato CM, et al. Social and medical needs of rare metabolic patients: results from a MetabERN survey. Orphanet J Rare Dis. 2021;16(1):336.
232. Servais A, Thomas K, Dello Strologo L, Sayer JA, Bekri S, Bertholet-Thomas A, et al. Cystinuria: clinical practice recommendation. Kidney Int. 2021;99(1):48–58.
233. Seppälä TT, Latchford A, Negoi I, Sampaio Soares A, Jimenez-Rodriguez R, Sánchez-Guillén L, et al. European guidelines from the EHTG and ESCP for Lynch syndrome: an updated third edition of the Mallorca guidelines based on gene and gender. Br J Surg. 2021;108(5):484–98.
234. Schneider DT, Orbach D, Ben-Ami T, Bien E, Bisogno G, Brecht IB, et al. Consensus recommendations from the EXPeRT/PARTNER groups for the diagnosis and therapy of sex cord stromal tumors in children and adolescents. Pediatr Blood Cancer. 2021;68(Suppl 4):e29017.
235. Reschke F, Rohayem J, Maffei P, Dassie F, Schwandt A, de Beaufort C, et al. Collaboration for rare diabetes: understanding new treatment options for Wolfram syndrome. Endocrine. 2021;71(3):626–33.
236. Raga S, Specchio N, Rheims S, Wilmshurst JM. Developmental and epileptic encephalopathies: recognition and approaches to care. Epileptic Disord. 2021;23(1):40–52.
237. Persani L, Bonomi M, Cools M, Dattani M, Dunkel L, Gravholt CH, et al. ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. Endocrine. 2021;71(3):681–8.
238. Nowotny H, Ahmed SF, Bensing S, Beun JG, Brösamle M, Chifu I, et al. Therapy options for adrenal insufficiency and recommendations for the management of adrenal crisis. Endocrine. 2021;71(3):586–94.
239. Northup PG, Garcia-Pagan JC, Garcia-Tsao G, Intagliata NM, Superina RA, Roberts LN, et al. Vascular Liver Disorders, Portal Vein Thrombosis, and Procedural Bleeding in Patients With Liver Disease: 2020 Practice Guidance by the American Association for the Study of Liver Diseases. Hepatology. 2021;73(1):366–413.
240. Marmolejo DH, Wong MYZ, Bajalica-Lagercrantz S, Tischkowitz M, Balmaña J. Overview of hereditary breast and ovarian cancer (HBOC) guidelines across Europe. Eur J Med Genet. 2021;64(12):104350.
241. Legius E, Messiaen L, Wolkenstein P, Pancza P, Avery RA, Berman Y, et al. Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genet Med. 2021;23(8):1506–13.
242. Kusamura S, Kepenekian V, Villeneuve L, Lurvink RJ, Govaerts K, De Hingh I, et al. Peritoneal mesothelioma: PSOGI/EURACAN clinical practice guidelines for diagnosis, treatment and follow-up. Eur J Surg Oncol. 2021;47(1):36–59.
243. Kridin K, Brüggen MC, Chua SL, Bygum A, Walsh S, Nägeli MC, et al. Assessment of Treatment Approaches and Outcomes in Stevens-Johnson Syndrome and Toxic Epidermal Necrolysis: Insights From a Pan-European Multicenter Study. JAMA Dermatol. 2021;157(10):1182–90.
244. Konrad M, Nijenhuis T, Ariceta G, Bertholet-Thomas A, Calo LA, Capasso G, et al. Diagnosis and management of Bartter syndrome: executive summary of the consensus and recommendations from the European Rare Kidney Disease Reference Network Working Group for Tubular Disorders. Kidney Int. 2021;99(2):324–35.
245. Iolascon A, Bianchi P, Andolfo I, Russo R, Barcellini W, Fermo E, et al. Recommendations for diagnosis and treatment of methemoglobinemia. Am J Hematol. 2021;96(12):1666–78.
246. Has C, El Hachem M, Bučková H, Fischer P, Friedová M, Greco C, et al. Practical management of epidermolysis bullosa: consensus clinical position statement from the European Reference Network for Rare Skin Diseases. J Eur Acad Dermatol Venereol. 2021;35(12):2349–60.
247. Gronchi A, Miah AB, Dei Tos AP, Abecassis N, Bajpai J, Bauer S, et al. Soft tissue and visceral sarcomas: ESMO-EURACAN-GENTURIS Clinical Practice Guidelines for diagnosis, treatment and follow-up(☆). Ann Oncol. 2021;32(11):1348–65.
248. Govaerts K, Lurvink RJ, De Hingh I, Van der Speeten K, Villeneuve L, Kusamura S, et al. Appendiceal tumours and pseudomyxoma peritonei: Literature review with PSOGI/EURACAN clinical practice guidelines for diagnosis and treatment. Eur J Surg Oncol. 2021;47(1):11–35.
249. Gladdy RA. Precision guidelines for soft tissue and visceral sarcomas: the evidence, expert experience and ensuring optimal care for rare cancers, a 2021 update from ESMO-EURACAN-GENTURIS. Ann Oncol. 2021;32(11):1325–6.
250. Garcia-Pavia P, Rapezzi C, Adler Y, Arad M, Basso C, Brucato A, et al. Diagnosis and treatment of cardiac amyloidosis. A position statement of the European Society of Cardiology Working Group on Myocardial and Pericardial Diseases. Eur J Heart Fail. 2021;23(4):512–26.
251. Garcia-Pavia P, Bengel F, Brito D, Damy T, Duca F, Dorbala S, et al. Expert consensus on the monitoring of transthyretin amyloid cardiomyopathy. Eur J Heart Fail. 2021;23(6):895–905.
252. Fleseriu M, Auchus R, Bancos I, Ben-Shlomo A, Bertherat J, Biermasz NR, et al. Consensus on diagnosis and management of Cushing's disease: a guideline update. Lancet Diabetes Endocrinol. 2021;9(12):847–75.
253. Ferrari A, Lopez Almaraz R, Reguerre Y, Cesen M, Bergamaschi L, Indini A, et al. Cutaneous melanoma in children and adolescents: The EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e28992.
254. Dingemann C, Eaton S, Aksnes G, Bagolan P, Cross KM, De Coppi P, et al. ERNICA Consensus Conference on the Management of Patients with Long-Gap Esophageal Atresia: Perioperative, Surgical, and Long-Term Management. Eur J Pediatr Surg. 2021;31(3):214–25.
255. Dhaenens BAE, Ferner RE, Bakker A, Nievo M, Evans DG, Wolkenstein P, et al. Identifying challenges in neurofibromatosis: a modified Delphi procedure. Eur J Hum Genet. 2021;29(11):1625–33.
256. de la Fouchardiere A, Blokx W, van Kempen LC, Luzar B, Piperno-Neumann S, Puig S, et al. ESP, EORTC, and EURACAN Expert Opinion: practical recommendations for the pathological diagnosis and clinical management of intermediate melanocytic tumors and rare related melanoma variants. Virchows Arch. 2021;479(1):3–11.
257. Boyer O, Schaefer F, Haffner D, Bockenhauer D, Hölttä T, Bérody S, et al. Management of congenital nephrotic syndrome: consensus recommendations of the ERKNet-ESPN Working Group. Nat Rev Nephrol. 2021;17(4):277–89.
258. Bouys L, Chiodini I, Arlt W, Reincke M, Bertherat J. Update on primary bilateral macronodular adrenal hyperplasia (PBMAH). Endocrine. 2021;71(3):595–603.
259. Bossi P, Chan AT, Licitra L, Trama A, Orlandi E, Hui EP, et al. Nasopharyngeal carcinoma: ESMO-EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up(†). Ann Oncol. 2021;32(4):452–65.
260. Bodemer C, Steijlen P, Mazereeuw-Hautier J, O'Toole EA. Treatment of hereditary palmoplantar keratoderma: a review by analysis of the literature. Br J Dermatol. 2021;184(3):393–400.
261. Bisogno G, Sarnacki S, Stachowicz-Stencel T, Minard Colin V, Ferrari A, Godzinski J, et al. Pleuropulmonary blastoma in children and adolescents: The EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29045.
262. Bien E, Roganovic J, Krawczyk MA, Godzinski J, Orbach D, Cecchetto G, et al. Pancreatoblastoma in children: EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29112.
263. Ben-Ami T, Kontny U, Surun A, Brecht IB, Almaraz RL, Dragomir M, et al. Nasopharyngeal carcinoma in children and adolescents: The EXPeRT/PARTNER diagnostic and therapeutic recommendations. Pediatr Blood Cancer. 2021;68(Suppl 4):e29018.
264. Baumgartner H, De Backer J, Babu-Narayan SV, Budts W, Chessa M, Diller GP, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. Eur Heart J. 2021;42(6):563–645.

265. Bakhuizen JJ, Hanson H, van der Tuin K, Lalloo F, Tischkowitz M, Wadt K, et al. Surveillance recommendations for DICER1 pathogenic variant carriers: a report from the SIOPE Host Genome Working Group and CanGene-CanVar Clinical Guideline Working Group. Fam Cancer. 2021;20(4):337–48.
266. Baker A, Frauca Remacha E, Torres Canizales J, Bravo-Gallego LY, Fitzpatrick E, Alonso Melgar A, et al. Current practices on diagnosis, prevention and treatment of post-transplant lymphoproliferative disorder in pediatric patients after solid organ transplantation: Results of ERN TransplantChild Healthcare Working Group Survey. Child (Basel). 2021;8(8).
267. Appelman-Dijkstra NM, Ertl DA, Zillikens MC, Rjenmark L, Winter EM. Hypercalcemia during pregnancy: management and outcomes for mother and child. Endocrine. 2021;71(3):604–10.
268. Andreou D. The ESMO-EURACAN-GENTURIS-ERNPaedCan Clinical Practice Guidelines for bone sarcomas - the challenges of the fight against rare cancers. Ann Oncol. 2021;32(12):1470–1.
269. Aleksovska K, Bassetti CLA, Berger T, Carvalho V, Costa J, Deuschl G, et al. Guidelines should be guidelines: Time to leave the terms consensus and position for other purposes. Eur J Neurol. 2021;28(8):2461–6.
270. van den Munckhof B, Arzimanoglou A, Perucca E, van Teeseling HC, Leijten FSS, Braun KPJ, et al. Corticosteroids versus clobazam in epileptic encephalopathy with ESES: a European multicentre randomised controlled clinical trial (RESCUE ESES*). Trials. 2020;21(1):957.
271. Tischkowitz M, Huang S, Banerjee S, Hague J, Hendricks WPD, Huntsman DG, et al. Small-Cell Carcinoma of the Ovary, Hypercalcemic Type-Genetics, New Treatment Targets, and Current Management Guidelines. Clin Cancer Res. 2020;26(15):3908–17.
272. Tischkowitz M, Colas C, Pouwels S, Hoogerbrugge N. Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. Eur J Hum Genet. 2020;28(10):1387–93.
273. Talarico R, Marinello D, Bombardieri S, Burmester G, Fonseca J, Frank C, et al. Clinical practice guidelines adherence, knowledge and awareness in rare and complex connective tissue diseases across Europe: results from the first ERN ReCONNET survey. RMD Open. 2020;6(2):0.
274. Tahani N, Maffei P, Dollfus H, Paisey R, Valverde D, Milan G, et al. Consensus clinical management guidelines for Alström syndrome. Orphanet J Rare Dis. 2020;15(1):253.
275. Schaaf J, Sedlmayr M, Prokosch HU, Ganslandt T, Schade-Brittinger C, von Wagner M, et al. The Status Quo of Rare Diseases Centres for the Development of a Clinical Decision Support System - A Cross-Sectional Study. Stud Health Technol Inf. 2020;271:176–83.
276. Nielsen JC, Lin YJ, de Oliveira Figueiredo MJ, Sepehri Shamloo A, Alfie A, Boveda S, et al. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) expert consensus on risk assessment in cardiac arrhythmias: use the right tool for the right outcome, in the right population. Heart Rhythm. 2020;17(9):e269–316.
277. Nicholson AG, Sauter JL, Nowak AK, Kindler HL, Gill RR, Remy-Jardin M, et al. EURACAN/IASLC Proposals for Updating the Histologic Classification of Pleural Mesothelioma: Towards a More Multidisciplinary Approach. J Thorac Oncol. 2020;15(1):29–49.
278. Monahan KJ, Bradshaw N, Dolwani S, Desouza B, Dunlop MG, East JE, et al. Guidelines for the management of hereditary colorectal cancer from the British Society of Gastroenterology (BSG)/Association of Coloproctology of Great Britain and Ireland (ACPGBI)/United Kingdom Cancer Genetics Group (UKCGG). Gut. 2020;69(3):411–44.
279. Mellerio JE, El Hachem M, Bellon N, Zambruno G, Buckova H, Autrata R, et al. Emergency management in epidermolysis bullosa: consensus clinical recommendations from the European reference network for rare skin diseases. Orphanet J Rare Dis. 2020;15(1):142.
280. Martin Saborido C, Borobia AM, Cobas J, D'Antiga L, Frauca E, Hernández-Oliveros F, et al. Effectiveness of immunosuppression minimisation, conversion or withdrawal strategies in paediatric solid organ and haematopoietic stem cell transplantation: a protocol of a systematic review and meta-analysis. BMJ Open. 2020;10(12):e037721.
281. Lipska-Ziętkiewicz BS, Ozaltin F, Hölttä T, Bockenhauer D, Bérody S, Levtchenko E, et al. Genetic aspects of congenital nephrotic syndrome: a consensus statement from the ERKNet-ESPN inherited glomerulopathy working group. Eur J Hum Genet. 2020;28(10):1368–78.
282. Kyrklund K, Sloots CEJ, de Blaauw I, Bjørnland K, Rolle U, Cavalieri D, et al. ERNICA guidelines for the management of rectosigmoid Hirschsprung's disease. Orphanet J Rare Dis. 2020;15(1):164.
283. Kinsler VA, Boccara O, Fraitag S, Torrelo A, Vabres P, Diociaiuti A. Mosaic abnormalities of the skin: review and guidelines from the European Reference Network for rare skin diseases. Br J Dermatol. 2020;182(3):552–63.
284. Johannsen TH, Andersson AM, Ahmed SF, de Rijke YB, Greaves RF, Hartmann MF, et al. Peptide hormone analysis in diagnosis and treatment of Differences of Sex Development: joint position paper of EU COST Action 'DSDnet' and European Reference Network on Rare Endocrine Conditions. Eur J Endocrinol. 2020;182(6):P1–15.
285. Heard JM, Vrinten C, Schlander M, Bellettato CM, van Lingen C, Scarpa M. Availability, accessibility and delivery to patients of the 28 orphan medicines approved by the European Medicine Agency for hereditary metabolic diseases in the MetabERN network. Orphanet J Rare Dis. 2020;15(1):3.
286. Frebourg T, Bajalica Lagercrantz S, Oliveira C, Magenheim R, Evans DG. Guidelines for the Li-Fraumeni and heritable TP53-related cancer syndromes. Eur J Hum Genet. 2020;28(10):1379–86.
287. Faughnan ME, Mager JJ, Hetts SW, Palda VA, Lang-Robertson K, Buscarini E, et al. Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Ann Intern Med. 2020;173(12):989–1001.
288. Fassnacht M, Assie G, Baudin E, Eisenhofer G, de la Fouchardiere C, Haak HR, et al. Adrenocortical carcinomas and malignant phaeochromocytomas: ESMO-EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol. 2020;31(11):1476–90.
289. Evans DG, Woodward ER, Bajalica-Lagercrantz S, Oliveira C, Frebourg T. Germline TP53 Testing in Breast Cancers: Why, When and How? Cancers. (Basel). 2020;12(12).
290. Eker OF, Boccardi E, Sure U, Patel MC, Alicante S, Alsafi A, et al. European Reference Network for Rare Vascular Diseases (VASCERN) position statement on cerebral screening in adults and children with hereditary haemorrhagic telangiectasia (HHT). Orphanet J Rare Dis. 2020;15(1):165.
291. Edouard T, Guillaume-Czitrom S, Bacchetta J, Sermet-Gaudelus I, Dugelay E, Martinez-Vinson C, et al. Guidelines for the management of children at risk of secondary bone fragility: Expert opinion of a French working group. Arch Pediatr. 2020;27(7):393–8.
292. Dingemann C, Eaton S, Aksnes G, Bagolan P, Cross KM, De Coppi P, et al. ERNICA Consensus Conference on the Management of Patients with Esophageal Atresia and Tracheoesophageal Fistula: Diagnostics, Preoperative, Operative, and Postoperative Management. Eur J Pediatr Surg. 2020;30(4):326–36.
293. Dingemann C, Eaton S, Aksnes G, Bagolan P, Cross KM, De Coppi P, et al. ERNICA Consensus Conference on the Management of Patients with Esophageal Atresia and Tracheoesophageal Fistula: Follow-up and Framework. Eur J Pediatr Surg. 2020;30(6):475–82.
294. De Backer J, Bondue A, Budts W, Evangelista A, Gallego P, Jondeau G, et al. Genetic counselling and testing in adults with congenital heart disease: A consensus document of the ESC Working Group of Grown-Up Congenital Heart Disease, the ESC Working Group on Aorta and Peripheral Vascular Disease and the European Society of Human Genetics. Eur J Prev Cardiol. 2020;27(13):1423–35.
295. Davidson AE, Hafford-Tear NJ, Dudakova L, Sadan AN, Pontikos N, Hardcastle AJ, et al. CUGC for posterior polymorphous corneal dystrophy (PPCD). Eur J Hum Genet. 2020;28(1):126–31.
296. Brodszki N, Frazer-Abel A, Grumach AS, Kirschfink M, Litzman J, Perez E, et al. European Society for Immunodeficiencies (ESID) and European Reference Network on Rare Primary Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA) Complement Guideline: Deficiencies, Diagnosis, and Management. J Clin Immunol. 2020;40(4):576–91.
297. Bodemer C, Diociaiuti A, Hadj-Rabia S, Robert MP, Desguerre I, Manière MC, et al. Multidisciplinary consensus recommendations from a European network for the diagnosis and practical management of patients with incontinentia pigmenti. J Eur Acad Dermatol Venereol. 2020;34(7):1415–24.
298. Blair VR, McLeod M, Carneiro F, Coit DG, D'Addario JL, van Dieren JM, et al. Hereditary diffuse gastric cancer: updated clinical practice guidelines. Lancet Oncol. 2020;21(8):e386–97.
299. Baldelli I, Baccarani A, Barone C, Bedeschi F, Bianca S, Calabrese O, et al. Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet J Rare Dis. 2020;15(1):201.
300. Atalaia A, Thompson R, Corvo A, Carmody L, Piscia D, Matalonga L, et al. A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet J Rare Dis. 2020;15(1):206.

301. Arzimanoglou A, Brandl U, Cross JH, Gil-Nagel A, Lagae L, Landmark CJ, et al. Epilepsy and cannabidiol: a guide to treatment. Epileptic Disord. 2020;22(1):1–14.
302. van Rijssen TJ, van Dijk EHC, Yzer S, Ohno-Matsui K, Keunen JEE, Schlingemann RO, et al. Central serous chorioretinopathy: Towards an evidence-based treatment guideline. Prog Retin Eye Res. 2019;73:100770.
303. van Leerdam ME, Roos VH, van Hooft JE, Dekker E, Jover R, Kaminski MF, et al. Endoscopic management of polyposis syndromes: European Society of Gastrointestinal Endoscopy (ESGE) Guideline. Endoscopy. 2019;51(9):877–95.
304. van Leerdam ME, Roos VH, van Hooft JE, Balaguer F, Dekker E, Kaminski MF, et al. Endoscopic management of Lynch syndrome and of familial risk of colorectal cancer: European Society of Gastrointestinal Endoscopy (ESGE) Guideline. Endoscopy. 2019;51(11):1082–93.
305. van de Laar I, Arbustini E, Loeys B, Björck E, Murphy L, Groenink M, et al. European reference network for rare vascular diseases (VASCERN) consensus statement for the screening and management of patients with pathogenic ACTA2 variants. Orphanet J Rare Dis. 2019;14(1):264.
306. Towbin JA, McKenna WJ, Abrams DJ, Ackerman MJ, Calkins H, Darrieux FCC, et al. 2019 HRS expert consensus statement on evaluation, risk stratification, and management of arrhythmogenic cardiomyopathy: Executive summary. Heart Rhythm. 2019;16(11):e373–407.
307. O'Mahony C, Akhtar MM, Anastasiou Z, Guttmann OP, Vriesendorp PA, Michels M, et al. Effectiveness of the 2014 European Society of Cardiology guideline on sudden cardiac death in hypertrophic cardiomyopathy: a systematic review and meta-analysis. Heart. 2019;105(8):623–31.
308. Haffner D, Emma F, Eastwood DM, Biosse Duplan M, Bacchetta J, Schnabel D, et al. Clinical practice recommendations for the diagnosis and management of X-linked hypophosphataemia. Nat Rev Nephrol. 2019;15(7):435–55.
309. Grimm MJ, Respondek G, Stamelou M, Arzberger T, Ferguson L, Gelpi E, et al. How to apply the movement disorder society criteria for diagnosis of progressive supranuclear palsy. Mov Disord. 2019;34(8):1228–32.
310. Franceschi E, Hofer S, Brandes AA, Frappaz D, Kortmann RD, Bromberg J, et al. EANO-EURACAN clinical practice guideline for diagnosis, treatment, and follow-up of post-pubertal and adult patients with medulloblastoma. Lancet Oncol. 2019;20(12):e715–28.
311. Fellmann F, van El CG, Charron P, Michaud K, Howard HC, Boers SN, et al. European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. Eur J Hum Genet. 2019;27(12):1763–73.
312. Donal E, Delgado V, Bucciarelli-Ducci C, Galli E, Haugaa KH, Charron P, et al. Multimodality imaging in the diagnosis, risk stratification, and management of patients with dilated cardiomyopathies: an expert consensus document from the European Association of Cardiovascular Imaging. Eur Heart J Cardiovasc Imaging. 2019;20(10):1075–93.
313. Crosbie EJ, Ryan NAJ, Arends MJ, Bosse T, Burn J, Cornes JM, et al. The Manchester International Consensus Group recommendations for the management of gynecological cancers in Lynch syndrome. Genet Med. 2019;21(10):2390–400.
314. Cheung MS, Alves I, Hagenäs L, Mohnike K. Meeting report from the achondroplasia foramen magnum workshop, Salzburg, Austria 22nd June 2019. Bone. 2019;127:499–502.
315. Bianchi P, Fermo E, Glader B, Kanno H, Agarwal A, Barcellini W, et al. Addressing the diagnostic gaps in pyruvate kinase deficiency: Consensus recommendations on the diagnosis of pyruvate kinase deficiency. Am J Hematol. 2019;94(1):149–61.
316. Bialer M, Cross H, Hedrich UBS, Lagae L, Lerche H, Loddenkemper T. Novel treatment approaches and pediatric research networks in status epilepticus. Epilepsy Behav. 2019;101:106564. Pt B).
317. Verhagen JMA, Kempers M, Cozijnsen L, Bouma BJ, Duijnhouwer AL, Post JG, et al. Expert consensus recommendations on the cardiogenetic care for patients with thoracic aortic disease and their first-degree relatives. Int J Cardiol. 2018;258:243–8.
318. Tamirou F, Arnaud L, Talarico R, Scirè CA, Alexander T, Amoura Z, et al. Systemic lupus erythematosus: state of the art on clinical practice guidelines. RMD Open. 2018;4(2):e000793.
319. Sulli A, Talarico R, Scirè CA, Avcin T, Castori M, Ferraris A, et al. Ehlers-Danlos syndromes: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000790.
320. Smith V, Scirè CA, Talarico R, Airo P, Alexander T, Allanore Y, et al. Systemic sclerosis: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000782.
321. Shovlin CL, Buscarini E, Kjeldsen AD, Mager HJ, Sabba C, Droege F, et al. European Reference Network For Rare Vascular Diseases (VASCERN) Outcome Measures For Hereditary Haemorrhagic Telangiectasia (HHT). Orphanet J Rare Dis. 2018;13(1):136.
322. Romão VC, Talarico R, Scirè CA, Vieira A, Alexander T, Baldini C, et al. Sjögren's syndrome: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000789.
323. Rednic S, Damian L, Talarico R, Scirè CA, Tobias A, Costedoat-Chalumeau N, et al. Relapsing polychondritis: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000788.
324. O'Mahony C, Jichi F, Ommen SR, Christiaans I, Arbustini E, Garcia-Pavia P, et al. International External Validation Study of the 2014 European Society of Cardiology Guidelines on Sudden Cardiac Death Prevention in Hypertrophic Cardiomyopathy (EVIDENCE-HCM). Circulation. 2018;137(10):1015–23.
325. Mosca M, Cutolo M. Clinical practice guidelines: the first year of activity of the European Reference Network on Rare and Complex Connective Tissue and Musculoskeletal Diseases (ERN ReCONNET). RMD Open. 2018;4(Suppl 1):e000791.
326. Meyer A, Scirè CA, Talarico R, Alexander T, Amoura Z, Avcin T, et al. Idiopathic inflammatory myopathies: state of the art on clinical practice guidelines [corrected]. RMD Open. 2018;4(Suppl 1):e000784.
327. Mantovani G, Bastepe M, Monk D, de Sanctis L, Thiele S, Usardi A, et al. Diagnosis and management of pseudohypoparathyroidism and related disorders: first international Consensus Statement. Nat Rev Endocrinol. 2018;14(8):476–500.
328. Lobitz S, Telfer P, Cela E, Allaf B, Angastiniotis M, Backman Johansson C, et al. Newborn screening for sickle cell disease in Europe: recommendations from a Pan-European Consensus Conference. Br J Haematol. 2018;183(4):648–60.
329. Limper M, Scirè CA, Talarico R, Amoura Z, Avcin T, Basile M, et al. Antiphospholipid syndrome: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000785.
330. Kölble K, Cross JH, Becker A, Blümcke I. A web-based diagnostic reference centre for the European Reference Network EpiCare: recommendations of the eNeuropathology working group. Epileptic Disord. 2018;20(5):339–45.
331. Iaccarino L, Talarico R, Scirè CA, Amoura Z, Burmester G, Doria A, et al. IgG4-related diseases: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000787.
332. Chaigne B, Scirè CA, Talarico R, Alexander T, Amoura Z, Avcin T, et al. Mixed connective tissue disease: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000783.
333. Castellani C, Duff AJA, Bell SC, Heijerman HGM, Munck A, Ratjen F, et al. ECFS best practice guidelines: the 2018 revision. J Cyst Fibros. 2018;17(2):153–78.
334. Casali PG, Bielack S, Abecassis N, Aro HT, Bauer S, Biagini R, et al. Bone sarcomas: ESMO-PaedCan-EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol. 2018;29(Suppl 4):iv79–95.
335. Casali PG, Abecassis N, Aro HT, Bauer S, Biagini R, Bielack S, et al. Gastrointestinal stromal tumours: ESMO-EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up. Ann Oncol. 2018;29(Suppl 4):iv267.
336. Cannizzo S, Lorenzoni V, Palla I, Pirri S, Trieste L, Triulzi I, et al. Rare diseases under different levels of economic analysis: current activities, challenges and perspectives. RMD Open. 2018;4(Suppl 1):e000794.
337. Antunes M, Scirè CA, Talarico R, Alexander T, Avcin T, Belocchi C, et al. Undifferentiated connective tissue disease: state of the art on clinical practice guidelines. RMD Open. 2018;4(Suppl 1):e000786.
338. Prades J, Kozhaeva O, Otth M, Kearns P, Ladenstein R, Rizzari C, et al. Do national cancer control plans address care and research for children, adolescents, and young adults? A review of status, priorities, and recommendations across 41 European countries. Lancet Reg Health Eur. 2025;49:101155.
339. Marrani E, Zajc Avramovic M, Marinello D, Talarico R, Baldini C, Collado-González E, et al. Challenges in the transition of care for rare connective tissue diseases: results from the 2023 ERN ReCONNET Transition of Care Task Force survey. Rheumatol Adv Pract. 2025;9(1):rkae149.
340. Kälviäinen R, Hadj-Allal Z, Kirjavainen J, Roivainen R, Linnankivi T, Peltola J, et al. Epilepsy care pathway: The Finnish model. Epilepsia Open. 2025;10(1):177–85.
341. Ingimarsdóttir IJ, Vishram-Nielsen JKK, Einarsson H, Goldfeder S, Mewton N, Barasa A, et al. Diagnostic and therapeutic practice for HFpEF across continents and regions: An international survey. ESC Heart Fail. 2025;12(1):487–96.
342. Arnaud L, Sander O, Rednic S, Mertz P, Faria R, Crisafulli F, et al. European Reference Network (ERN) ReCONNET methodology for the cross-cultural adaptation of instruments for research and care in the context of rare connective tissue diseases (CROSSADAPT). Orphanet J Rare Dis. 2025;20(1):230.

343. Wlodkowski T, Haeberle S, Schaefer F. [The European Rare Kidney Disease Reference Network]. Inn Med (Heidelb). 2024;65(12):1283–92.
344. Voi V, Gutierrez-Valle V, Cuzzubbo D, McMahon C, Casale M, Mañú Pereira MDM, et al. Limited access to transcranial Doppler screening and stroke prevention for children with sickle cell disease in Europe: Results of a multinational EuroBloodNet survey. Pediatr Blood Cancer. 2024;71(10):e31190.
345. Verdonschot JAJ, Heymans SRB, Van Linthout S. The next era of gene-specific clinical care in patients with dilated cardiomyopathy. Eur J Heart Fail. 2024;26(10):2169–72.
346. Talarico R, Marinello D, Palla I, Cannizzo S, Galetti I, Farrington S, et al. Improving organisation to improve care: ERN ReCONNET organisational reference model for systemic sclerosis patients' care pathway. J Scleroderma Relat Disord. 2024:23971983241269109.
347. Regnier M, Flammier S, Boutaba M, Ndongo AA, Servais A, Schaefer F, et al. Worldwide disparities in access to treatment and investigations for nephropathic cystinosis: a 2023 perspective. Pediatr Nephrol. 2024;39(4):1113–23.
348. Iannotta M, d'Aiello FA, Van De Bruaene A, Caruso R, Conte G, Ferrero P, et al. Modern tools in congenital heart disease imaging and procedure planning: a European survey. J Cardiovasc Med (Hagerstown). 2024;25(1):76–87.
349. Auvin S, Arzimanoglou A, Brambilla I, French J, Knupp KG, Lagae L, et al. Call for the use of the ILAE terminology for seizures and epilepsies by health care professionals and regulatory agencies to benefit patients and caregivers. Epilepsia. 2024;65(2):283–6.
350. Wilde AAM, Schwartz PJ, Long QT. Syndrome, a Diagnosis That Warrants Expert Opinion and Expert Centers. J Am Coll Cardiol. 2023;81(5):487–9.
351. Vetti HH, Blanco I, Menko FH. European collaboration on genetic tumour risk syndromes. Eur J Med Genet. 2023;66(2):104691.
352. Mañú Pereira MDM, Colombatti R, Alvarez F, Bartolucci P, Bento C, Brunetta AL, et al. Sickle cell disease landscape and challenges in the EU: the ERN-EuroBloodNet perspective. Lancet Haematol. 2023;10(8):e687–94.
353. Humbert M, Wagner T. The European reference network on rare lung diseases (ERN-LUNG) ambition to offer better lung health for all. Presse Med. 2023;52(3):104172.
354. Donà D, Bravo-Gallego LY, Remacha EF, Cananzi M, Gastaldi A, Canizalez JT, et al. Vaccination practices in pediatric transplantation: A survey among member centers of the European reference network TransplantChild. Pediatr Transpl. 2023;27(7):e14589.
355. Adachi T, El-Hattab AW, Jain R, Nogales Crespo KA, Quirland Lazo CI, Scarpa M, et al. Enhancing equitable access to rare disease diagnosis and treatment around the world: a review of evidence, policies, and challenges. Int J Environ Res Public Health. 2023;20(6).
356. Sikonja J, Groselj U, Scarpa M, la Marca G, Cheillan D, Kölker S, et al. Towards achieving equity and innovation in newborn screening across Europe. Int J Neonatal Screen. 2022;8(2).
357. Scarpa M, Bonham JR, Dionisi-Vici C, Prevot J, Pergent M, Meyts I, et al. Newborn screening as a fully integrated system to stimulate equity in neonatal screening in Europe. Lancet Reg Health Eur. 2022;13:100311.
358. Oomen L, Leijte E, Shilhan DE, Battye M, Feitz WFJ. Rare and Complex Urology: Clinical Overview of ERN eUROGEN. Eur Urol. 2022;81(2):204–12.
359. Junge N, Di Giorgio A, Girard M, Demir Z, Kaminska D, Janowska M, et al. Cold ischemia time and graft fibrosis are associated with autoantibodies after pediatric liver transplantation: a retrospective cohort study of the european reference network TransplantChild. Child (Basel). 2022;9(2).
360. Jondeau G, Hallac J, Hurard M, Daoud K, Barr N, Domnez I. The VASCERN European Reference Network: An overview. Eur J Med Genet. 2022;65(3):104420.
361. Behr ER, Scrocco C, Wilde AAM, Marijon E, Crotti L, Iliodromitis KE, et al. Investigation on Sudden Unexpected Death in the Young (SUDY) in Europe: results of the European Heart Rhythm Association Survey. Europace. 2022;24(2):331–9.
362. Amin AS, Biller R, Charron P, Wilde AAM. European Reference Network for rare, low prevalence, or complex diseases of the heart (ERN GUARD-Heart): 5 year anniversary. Eur Heart J. 2022;43(23):2173–6.
363. Schlegelberger B, Mecucci C, Wlodarski M. Review of guidelines for the identification and clinical care of patients with genetic predisposition for hematological malignancies. Fam Cancer. 2021;20(4):295–303.
364. Markiewicz-Kijewska M, Kaliciński P, Torres Canizales J, Di Giorgio A, Baumann U, Jorns C, et al. ABO Incompatible Liver Transplantation in Children: A 20 Year Experience from Centres in the TransplantChild European Reference Network. Child (Basel). 2021;8(9).
365. Laue T, Demir Z, Debray D, Cananzi M, Gaio P, Casotti V, et al. Undervaccination in pediatric liver transplant candidates with acute and chronic liver disease-a retrospective observational study of the European reference network TransplantChild. Child (Basel). 2021;8(8).
366. Grammens J, Schechter MY, Desender L, Claeys T, Sinatti C, VandeWalle J, et al. Pediatric Challenges in Robot-Assisted Kidney Transplantation. Front Surg. 2021;8:649418.
367. Ferrari A, Schneider DT, Bisogno G, Reguerre Y, Godzinski J, Bien E, et al. Facing the challenges of very rare tumors of pediatric age: The European Cooperative Study Group for Pediatric Rare Tumors (EXPeRT) background, goals, and achievements. Pediatr Blood Cancer. 2021;68(Suppl 4):e28993.
368. Conte G, Wilde A, Behr ER, Scherr D, Lenarczyk R, Gandjbachkh E, et al. Importance of Dedicated Units for the Management of Patients With Inherited Arrhythmia Syndromes. Circ Genom Precis Med. 2021;14(2):e003313.
369. Black GC, Sergouniotis P, Sodi A, Leroy BP, Van Cauwenbergh C, Liskova P, et al. The need for widely available genomic testing in rare eye diseases: an ERN-EYE position statement. Orphanet J Rare Dis. 2021;16(1):142.
370. Baumgartner T, Carreño M, Rocamora R, Bisulli F, Boni A, Brázdil M, et al. A survey of the European Reference Network EpiCARE on clinical practice for selected rare epilepsies. Epilepsia Open. 2021;6(1):160–70.
371. Jara P, Baker A, Baumann U, Borobia AM, Branchereu S, Candusso M, et al. Cross-cutting view of current challenges in paediatric solid organ and haematopoietic stem cell transplantation in Europe: the European Reference Network TransplantChild. Orphanet J Rare Dis. 2020;15(1):16.
372. Pitkänen A, Henshall DC, Cross JH, Guerrini R, Jozwiak S, Kokaia M, et al. Advancing research toward faster diagnosis, better treatment, and end of stigma in epilepsy. Epilepsia. 2019;60(7):1281–92.
373. Schaefer J, Tegtbauer N, Pfeiffer W, Wagner TOF, Storf H. Mapping of Health Care Providers for People with Rare Diseases - From Vision to Implementation. Stud Health Technol Inf. 2018;247:940–4.
374. Babac A, Frank M, Pauer F, Litzkendorf S, Rosenfeldt D, Lührs V, et al. Telephone health services in the field of rare diseases: a qualitative interview study examining the needs of patients, relatives, and health care professionals in Germany. BMC Health Serv Res. 2018;18(1):99.
375. Velasco Puyo P, Christou S, Campisi S, Rodríguez-Sánchez MA, Reidel S, Perez-Hoyo S, et al. COVID- 19 in patients affected by red blood cell disorders, results from the European registry ERN-EuroBloodNet. Orphanet J Rare Dis. 2025;20(1):183.
376. Murray SM, Pose E, Wittner M, Londoño MC, Schaub G, Cook J, et al. Immune responses and clinical outcomes after COVID-19 vaccination in patients with liver disease and liver transplant recipients. J Hepatol. 2024;80(1):109–23.
377. Karbiener M, Kindle G, Meyts I, Seppänen MRJ, Candotti F, Kamieniak M, et al. Clinical efficacy of SARS-CoV-2 Omicron-neutralizing antibodies in immunoglobulin preparations for the treatment of agammaglobulinemia in patients with primary antibody deficiency. J Med Virol. 2024;96(6):e29738.
378. Aubert O, Gosemann JH, Wijnen R, Lacher M, Ure B, Madadi-Sanjani O. Impact of the COVID-19 Pandemic on Pediatric Surgical Scientific Formats: An ERNICA Survey on Challenges and Future Directions. Eur J Pediatr Surg. 2024;34(3):267–73.
379. Tani C, Cardelli C, Depascale R, Gamba A, Iaccarino L, Doria A, et al. Long-term outcomes of COVID-19 vaccination in patients with rare and complex connective tissue diseases: The ERN-ReCONNET VACCINATE study. J Transl Autoimmun. 2023;7:100221.
380. Talarico R, Ramirez GA, Barreira SC, Cardamone C, Triggianese P, Aguilera S, et al. ERN ReCONNET points to consider for treating patients living with autoimmune rheumatic diseases with antiviral therapies and anti-SARS-CoV-2 antibody products. Clin Exp Rheumatol. 2023;41(3):543–53.
381. Salek M, Mueller A, Alanbousi I, Cepowska Z, Dutkiewicz M, Earl J, et al. Development of a centralised triage centre for children with cancer and blood disorders in response to the humanitarian crisis in Ukraine. Lancet Oncol. 2023;24(12):1315–8.
382. Nowotny HF, Bryce J, Ali SR, Giordano R, Baronio F, Chifu I, et al. Outcome of COVID-19 infections in patients with adrenal insufficiency and excess. Endocr Connect. 2023;12(4).
383. Bonuccelli A, Depietri G, Baldaccini T, Ricciutelli I, Peroni D, Spalice A, et al. Essential headaches in developmental age: What is changed before, during and after the lockdown for COVID-19 pandemic. Clinical study. Front Pediatr. 2023;11:1166984.
384. Zhou Y, Shalhoub R, Rogers SN, Yu S, Gu M, Fabre MA, et al. Clonal hematopoiesis is not significantly associated with COVID-19 disease severity. Blood. 2022;140(14):1650–5.
385. Talarico R, Aguilera S, Alexander T, Amoura Z, Antunes AM, Arnaud L, et al. Publisher Correction: The impact of COVID-19 on rare and complex

connective tissue diseases: the experience of ERN ReCONNET. Nat Rev Rheumatol. 2022;18(12):734.
386. Paneghetti L, Bellettato CM, Sechi A, Stepien KM, Scarpa M. One year of COVID-19: infection rates and symptoms in patients with inherited metabolic diseases followed by MetabERN. Orphanet J Rare Dis. 2022;17(1):109.
387. Muccioli L, Zenesini C, Taruffi L, Licchetta L, Mostacci B, Di Vito L, et al. Risk of hospitalization and death for COVID-19 in persons with epilepsy over a 20-month period: The EpiLink Bologna cohort, Italy. Epilepsia. 2022;63(9):2279–89.
388. Manry J, Bastard P, Gervais A, Le Voyer T, Rosain J, Philippot Q, et al. The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proc Natl Acad Sci U S A. 2022;119(21):e2200413119.
389. Juul N, Cazals A, Hofmann A, Amesty V, Verkauskas G, Dobrowolska-Glazar B, et al. How the First Year of COVID-19 Affected Elective Pediatric Urology Patients: A Longitudinal Study Based on Waiting Lists and Surveys From 10 European Centers. Front Public Health. 2022;10:874758.
390. Dollfus H, Arzimanoglou A, Scarpa M. The European Reference Networks for rare and complex diseases respond to the Ukrainian crisis. Lancet Reg Health - Europe. 2022;19.
391. Degenhardt F, Ellinghaus D, Juzenas S, Lerga-Jaso J, Wendorff M, Maya-Miles D, et al. Detailed stratified GWAS analysis for severe COVID-19 in four European populations. Hum Mol Genet. 2022;31(23):3945–66.
392. Asselta R, Paraboschi EM, Stravalaci M, Invernizzi P, Bonfanti P, Biondi A, et al. Reply to: Hultström Genetic determinants of mannose-binding lectin activity predispose to thromboembolic complications in critical COVID-19. Mannose-binding lectin genetics in COVID-19. Nat Immunol. 2022;23(6):865–7.
393. Asadi-Pooya AA, Trinka E, Hingray C, Fawaz A, Karakis I, Kishk NA, et al. An international study of the effects of the COVID-19 pandemic on characteristics of functional seizures. Epilepsy Behav. 2022;127:108530.
394. Agulnik A, Kizyma R, Salek M, Wlodarski MW, Pogorelyy M, Oszer A, et al. Global effort to evacuate Ukrainian children with cancer and blood disorders who have been affected by war. Lancet Haematol. 2022;9(9):e645–7.
395. ESC guidance for the. diagnosis and management of cardiovascular disease during the COVID-19 pandemic: part 2-care pathways, treatment, and follow-up. Eur Heart J. 2022;43(11):1059–103.
396. European Society of Cardiology guidance for the. diagnosis and management of cardiovascular disease during the COVID-19 pandemic: part 1-epidemiology, pathophysiology, and diagnosis. Eur Heart J. 2022;43(11):1033–58.
397. Sangiorgi L, Brizola E. The line between COVID-19 pandemic and rare bone diseases. Ir J Med Sci. 2021;190(3):1243–4.
398. Sanchez-Larsen A, Conde-Blanco E, Viloria-Alebesque A, Sánchez-Vizcaíno Buendía C, Espinosa Oltra T, Alvarez-Noval A, et al. COVID-19 prevalence and mortality in people with epilepsy: A nation-wide multicenter study. Epilepsy Behav. 2021;125:108379.
399. Pisella LI, Fernandes S, Solé G, Stojkovic T, Tard C, Chanson JB, et al. A multicenter cross-sectional French study of the impact of COVID-19 on neuromuscular diseases. Orphanet J Rare Dis. 2021;16(1):450.
400. Marlais M, Wlodkowski T, Al-Akash S, Ananin P, Bandi VK, Baudouin V, et al. COVID-19 in children treated with immunosuppressive medication for kidney diseases. Arch Dis Child. 2021;106(8):798–801.
401. De Groot CJ, Poitou Bernert C, Coupaye M, Clement K, Paschou SA, Charmandari E, et al. Clinical management of patients with genetic obesity during COVID-19 pandemic: position paper of the ESE Growth & Genetic Obesity COVID-19 Study Group and Rare Endo-ERN main thematic group on Growth and Obesity. Endocrine. 2021;71(3):653–62.
402. Cross JH, Kwon CS, Asadi-Pooya AA, Balagura G, Gómez-Iglesias P, Guekht A, et al. Epilepsy care during the COVID-19 pandemic. Epilepsia. 2021;62(10):2322–32.
403. Cakir OO, Castiglione F, Tandogdu Z, Collins J, Alnajjar HM, Akers C, et al. Management of penile cancer patients during the COVID-19 pandemic: An eUROGEN accelerated Delphi consensus study. Urol Oncol. 2021;39(3):e1979–17.
404. Brambilla I, Aibar J, Hallet AS, Bibic I, Cardenal-Muñoz E, Prpic I, et al. Impact of the COVID-19 lockdown on patients and families with Dravet syndrome. Epilepsia Open. 2021;6(1):216–24.
405. Beniczky S, Husain A, Ikeda A, Alabri H, Helen Cross J, Wilmshurst J, et al. Importance of access to epilepsy monitoring units during the COVID-19 pandemic: Consensus statement of the International League against epilepsy and the International Federation of Clinical Neurophysiology. Clin Neurophysiol. 2021;132(9):2248–50.
406. Wirrell EC, Grinspan ZM, Knupp KG, Jiang Y, Hammeed B, Mytinger JR, et al. Care Delivery for Children With Epilepsy During the COVID-19 Pandemic: An International Survey of Clinicians. J Child Neurol. 2020;35(13):924–33.
407. Muccioli L, Pensato U, Cani I, Guerra L, Provini F, Bordin G, et al. COVID-19-related encephalopathy presenting with aphasia resolving following tocilizumab treatment. J Neuroimmunol. 2020;349:577400.
408. Mostacci B, Licchetta L, Cacciavillani C, Di Vito L, Ferri L, Menghi V, et al. The Impact of the COVID-19 Pandemic on People With Epilepsy. An Italian Survey and a Global Perspective. Front Neurol. 2020;11:613719.
409. Marlais M, Wlodkowski T, Vivarelli M, Pape L, Tönshoff B, Schaefer F, et al. The severity of COVID-19 in children on immunosuppressive medication. Lancet Child Adolesc Health. 2020;4(7):e17–8.
410. Lampe C, Dionisi-Vici C, Bellettato CM, Paneghetti L, van Lingen C, Bond S, et al. The impact of COVID-19 on rare metabolic patients and healthcare providers: results from two MetabERN surveys. Orphanet J Rare Dis. 2020;15(1):341.
411. Krysl D, Beniczky S, Franceschetti S, Arzimanoglou A. The COVID-19 outbreak and approaches to performing EEG in Europe. Epileptic Disord. 2020;22(5):548–54.
412. Granata T, Bisulli F, Arzimanoglou A, Rocamora R. Did the COVID-19 pandemic silence the needs of people with epilepsy? Epileptic Disord. 2020;22(4):439–42.
413. French JA, Brodie MJ, Caraballo R, Devinsky O, Ding D, Jehi L, et al. Keeping people with epilepsy safe during the COVID-19 pandemic. Neurology. 2020;94(23):1032–7.
414. Doná D, Torres Canizales J, Benetti E, Cananzi M, De Corti F, Calore E, et al. Pediatric transplantation in Europe during the COVID-19 pandemic: Early impact on activity and healthcare. Clin Transpl. 2020;34(10):e14063.
415. Brizola E, Adami G, Baroncelli GI, Bedeschi MF, Berardi P, Boero S, et al. Providing high-quality care remotely to patients with rare bone diseases during COVID-19 pandemic. Orphanet J Rare Dis. 2020;15(1):228.
416. Vujović A, Sellier-Leclerc AL, Mancuso MC, Boyer O, Awan A, Gargiulo A, et al. Real-world use of complement inhibitors for haemolytic uraemic syndrome: an analysis of the European Rare Kidney Disease Registry cohort. EClinicalMedicine. 2025;82:103159.
417. Torchia A, Vari S, Onesti CE, Ceddia S, Maschio M, Maggi G, et al. A narrative review on diagnosis and treatment of ifosfamide-induced encephalopathy, the perspective of a EURACAN reference center for sarcomas. Front Pharmacol. 2025;16:1512966.
418. Rossi D, Granström AL, Pakarinen M, Bjørnland K, de Blaauw I, Ellebæk M, et al. Defining Excellence: The First Core Set of Quality Indicators for the European Paediatric Surgical Audit on Hirschsprung's Disease Care. Acta Paediatr. 2025;114(6):1464–77.
419. Roganovic J, Virgone C, Ben-Ami T, Reguerre Y, Ferrari A, Orbach D, et al. Paediatric very rare tumours registration and management in European countries with low health expenditure average rates. Clin Transl Oncol. 2025;27(4):1779–88.
420. Gimpel C, Fieuws S, Hofstetter J, Pitcher D, Vanmeerbeek L, Haeberle S, et al. Insights from ADPedKD, ERKReg and RaDaR registries provide a multinational perspective on the presentation of childhood autosomal dominant polycystic kidney disease in high- and middle-income countries. Kidney Int. 2025;108(1):105–18.
421. Teunissen NM, Brendel J, Heurn L, Ure B, Wijnen R, Eaton S. Selection of Quality Indicators to Evaluate Quality of Care for Patients with Esophageal Atresia Using a Delphi Method. Eur J Pediatr Surg. 2024;34(5):398–409.
422. Schregel I, Papp M, Sipeki N, Kovats PJ, Taubert R, Engel B, et al. Unmet needs in autoimmune hepatitis: Results of the prospective multicentre European Reference Network Registry (R-LIVER). Liver Int. 2024;44(10):2687–99.
423. Schoenmakers DH, van den Berg S, Timmers L, Adang LA, Bäumer T, Bosch A, et al. Framework for Multistakeholder Patient Registries in the Field of Rare Diseases: Focus on Neurogenetic Diseases. Neurology. 2024;103(6):e209743.
424. Sanchez Gonzalez MDC, Kamerling P, Iermito M, Casati S, Riaz U, Veal CD, et al. Common conditions of use elements. Atomic concepts for consistent and effective information governance. Sci Data. 2024;11(1):465.
425. Priego Zurita AL, Boarini M, Casareto L, Cherenko M, Mordenti M, Moroni A, et al. The Role of the European Reference Network for Rare Bone Diseases (ERN BOND) and European Registries for Rare Bone and Mineral Conditions (EuRR-Bone) in the Governance of the Management of Rare Bone and Mineral Diseases. Calcif Tissue Int. 2024;115(5):498–506.
426. Oomen L, Bootsma-Robroeks C, Bouts AHM, Carbonell Pradas M, Gander R, Kienzl-Wagner K, et al. Pediatric kidney transplantation in Europe, a clinical snapshot pilot. Front Pediatr. 2024;12:1432027.

427. Jeanson F, Gibson SJ, Alper P, Bernier A, Woolley JP, Mietchen D, et al. Getting your DUCs in a row - standardising the representation of Digital Use Conditions. Sci Data. 2024;11(1):464.
428. Di Carlo D, Ladenstein R, Graf N, Merks JH, Hernández-Peñaloza G, Kearns P, et al. Common core variables for childhood cancer data integration. EJC Pediatr Oncol. 2024;4.
429. Cherenko M, Appelman-Dijkstra NM, Priego Zurita AL, Biermasz NR, Dekkers OM, Klok FA, et al. Venous thromboembolism in Cushing syndrome: results from an EuRRECa and Endo-ERN survey. Endocr Connect. 2024;13(6).
430. Atalaia A, Wandrei D, Lalout N, Thompson R, Tassoni A, t Hoen PAC, et al. EURO-NMD registry: federated FAIR infrastructure, innovative technologies and concepts of a patient-centred registry for rare neuromuscular disorders. Orphanet J Rare Dis. 2024;19(1):66.
431. Vyzhga Y, Hentgen V, Caorsi R, Wittkowski H, Hofer M, Ruperto N, et al. Breaking down the fences among registries on autoinflammatory diseases: the E-Merge project. Orphanet J Rare Dis. 2023;18(1):191.
432. Vergote S, De Bie FR, Duffy JMN, Bosteels J, Benachi A, Power B, et al. Core outcome set for perinatal interventions for congenital diaphragmatic hernia. Ultrasound Obstet Gynecol. 2023;62(3):374–82.
433. Varesio C, De Giorgis V, Veggiotti P, Nardocci N, Granata T, Ragona F, et al. GLUT1-DS Italian registry: past, present, and future: a useful tool for rare disorders. Orphanet J Rare Dis. 2023;18(1):63.
434. Trama A, Licitra L, Cavalieri S, Bonfarnuzzo S, Baili P, Ciarfella A, et al. The observational clinical registry (cohort design) of the European Reference Network on Rare Adult Solid Cancers: The protocol for the rare head and neck cancers. PLoS ONE. 2023;18(3):e0283071.
435. Teunissen N, Brendel J, Eaton S, Hall N, Thursfield R, van Heurn ELW, et al. Variability in the Reporting of Baseline Characteristics, Treatment, and Outcomes in Esophageal Atresia Publications: A Systematic Review. Eur J Pediatr Surg. 2023;33(2):129–37.
436. Priego Zurita AL, Grasemann C, Boarini M, Chapurlat R, Mordenti M, Javaid MK, et al. Data collection on rare bone and mineral conditions in Europe: The landscape of registries and databases. Eur J Med Genet. 2023;66(12):104868.
437. Mossey PA, Lai J, Meazzini MC, Breugem C, Mark H, van der Mink AB, et al. Core outcomes for orofacial clefts: reconciling traditional and ICHOM minimum datasets. Eur J Orthod. 2023;45(6):671–9.
438. Crotti L, Spazzolini C, Nyegaard M, Overgaard MT, Kotta MC, Dagradi F, et al. Clinical presentation of calmodulin mutations: the International Calmodulinopathy Registry. Eur Heart J. 2023;44(35):3357–70.
439. Collado A, Boaro MP, van der Veen S, Idrizovic A, Biemond BJ, Beneitez Pastor D, et al. Challenges and Opportunities of Precision Medicine in Sickle Cell Disease: Novel European Approach by GenoMed4All Consortium and ERN-EuroBloodNet. Hemasphere. 2023;7(3):e844.
440. Clement SC, Visser WE, Lebbink CA, Albano D, Claahsen-van der Grinten HL, Czarniecka A, et al. Development of a pediatric differentiated thyroid carcinoma registry within the EuRRECa project: rationale and protocol. Endocr Connect. 2023;12(3).
441. Balestrini S, Doccini V, Giometto S, Lucenteforte E, De Masi S, Giarola E, et al. A registry for Dravet syndrome: The Italian experience. Epilepsia Open. 2023;8(2):517–34.
442. Ali SR, Bryce J, Priego-Zurita AL, Cherenko M, Smythe C, de Rooij TM, et al. Electronic reporting of rare endocrine conditions within a clinical network: results from the EuRRECa project. Endocr Connect. 2023;12(12).
443. Schoenmakers DH, Beerepoot S, van den Berg S, Adang L, Bley A, Boelens JJ, et al. Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). Orphanet J Rare Dis. 2022;17(1):48.
444. Schaaf J, Zieschank A, Goebel J, Wagner TOF, Pennekamp P, Omran H, et al. The ERN-LUNG Population Registry: Aims, Software-Implementation and First Results. Stud Health Technol Inf. 2022;295:55–8.
445. Baumann U, Karam V, Adam R, Fondevila C, Dhawan A, Sokal E, et al. Prognosis of children undergoing liver transplantation: a 30-Year European Study. Pediatrics. 2022;150(4).
446. Bandeira M, Di Cianni F, Marinello D, Arnaud L, Cannizzo S, Carta C, et al. An overlook on the current registries for rare and complex connective tissue diseases and the future scenario of TogethERN ReCONNET. Front Med (Lausanne). 2022;9:889997.
447. Schaaf J, Chalmers J, Omran H, Pennekamp P, Sitbon O, Wagner TOF, et al. The Registry Data Warehouse in the European Reference Network for Rare Respiratory Diseases - Background, Conception and Implementation. Stud Health Technol Inf. 2021;278:41–8.
448. Sauter M, Belousova E, Benedik MP, Carter T, Cottin V, Curatolo P, et al. Rare manifestations and malignancies in tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increAse disease awareness (TOSCA). Orphanet J Rare Dis. 2021;16(1):301.
449. Rascon J, Salasevicius L, Rutkauskiene G, Bien E, Vincerzevskiene I. The impact of incomplete registration on survival rate of children with very rare tumors. Sci Rep. 2021;11(1):14066.
450. Orbach D, Ferrari A, Schneider DT, Reguerre Y, Godzinski J, Bien E, et al. The European Paediatric Rare Tumours Network - European Registry (PARTNER) project for very rare tumors in children. Pediatr Blood Cancer. 2021;68(Suppl 4):e29072.
451. Opladen T, Gleich F, Kozich V, Scarpa M, Martinelli D, Schaefer F, et al. U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet J Rare Dis. 2021;16(1):95.
452. Javaid MK, Mordenti M, Boarini M, Sangiorgi L, Westerheim I, Alves I, et al. Patients' priorities and expectations on an EU registry for rare bone and mineral conditions. Orphanet J Rare Dis. 2021;16(1):463.
453. Berger A, Rustemeier AK, Göbel J, Kadioglu D, Britz V, Schubert K, et al. How to design a registry for undiagnosed patients in the framework of rare disease diagnosis: suggestions on software, data set and coding system. Orphanet J Rare Dis. 2021;16(1):198.
454. Bassanese G, Wlodkowski T, Servais A, Heidet L, Roccatello D, Emma F, et al. The European Rare Kidney Disease Registry (ERKReg): objectives, design and initial results. Orphanet J Rare Dis. 2021;16(1):251.
455. Ali SR, Bryce J, Smythe C, Hytiris M, Priego AL, Appelman-Dijkstra NM, et al. Supporting international networks through platforms for standardised data collection-the European Registries for Rare Endocrine Conditions (EuRRECa) model. Endocrine. 2021;71(3):555–60.
456. Papa R, Cant A, Klein C, Little MA, Wulffraat NM, Gattorno M, et al. Towards European harmonisation of healthcare for patients with rare immune disorders: outcome from the ERN RITA registries survey. Orphanet J Rare Dis. 2020;15(1):33.
457. Morfouace M, Stevovic A, Vinches M, Golfinopoulos V, Jin DX, Holmes O, et al. First results of the EORTC-SPECTA/Arcagen study exploring the genomics of rare cancers in collaboration with the European reference network EURACAN. ESMO Open. 2020;5(6):e001075.
458. Sergouniotis PI, Maxime E, Leroux D, Olry A, Thompson R, Rath A, et al. An ontological foundation for ocular phenotypes and rare eye diseases. Orphanet J Rare Dis. 2019;14(1):8.
459. Köhler S, Carmody L, Vasilevsky N, Jacobsen JOB, Danis D, Gourdine JP, et al. Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Res. 2019;47(D1):D1018–27.
460. Crotti L, Spazzolini C, Tester DJ, Ghidoni A, Baruteau AE, Beckmann BM, et al. Calmodulin mutations and life-threatening cardiac arrhythmias: insights from the International Calmodulinopathy Registry. Eur Heart J. 2019;40(35):2964–75.
461. Ali SR, Bryce J, Cools M, Korbonits M, Beun JG, Taruscio D, et al. The current landscape of European registries for rare endocrine conditions. Eur J Endocrinol. 2019;180(1):89–98.
462. Thompson R, Robertson A, Lochmüller H. Natural History, Trial Readiness and Gene Discovery: Advances in Patient Registries for Neuromuscular Disease. Adv Exp Med Biol. 2017;1031:97–124.
463. Dos Santos Vieira B, Bernabe CH, Zhang S, Abaza H, Benis N, Camara A, et al. Towards FAIRification of sensitive and fragmented rare disease patient data: challenges and solutions in European reference network registries. Orphanet J Rare Dis. 2022;17(1):436.
464. Abaza H, Kadioglu D, Martin S, Papadopoulou A, Dos Santos Vieira B, Schaefer F, et al. Domain-Specific Common Data Elements for Rare Disease Registration: Conceptual Approach of a European Joint Initiative Toward Semantic Interoperability in Rare Disease Research. JMIR Med Inf. 2022;10(5):e32158.
465. Spivack OKC, Irvine WFE, Husby S, Representatives E, Wester T, Wijnen RMH. Fostering continuous quality improvement in a European rare disease network. Front Health Serv. 2025;5:1609018.

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