### RESEARCH



# Pregnancy-related issues in rare and low-prevalence diseases: results of ERN transversal working group on pregnancy and family planning survey



Dina Zucchi<sup>1,2†</sup>, Diana Marinello<sup>1†</sup>, Chiara Tani<sup>1</sup>, Giovanni Fulvio<sup>1</sup>, Silvia Aguilera<sup>3</sup>, Alexandra Benachi<sup>4</sup>, Ruth Biller<sup>5,6</sup>, Ignacio Blanco<sup>7</sup>, Petra Borgards<sup>8</sup>, Marie-Claude Boiteux<sup>9</sup>, Maria Luisa Brandi<sup>10</sup>, Ester Costafreda<sup>11</sup>, Joao E. Fonseca<sup>12,13</sup>, Micaela Fredi<sup>14</sup>, Violeta lotova<sup>15</sup>, Simone Louisse<sup>5</sup>, Cecilia Nalli<sup>14</sup>, Michela Onali<sup>16</sup>, Beverley Power<sup>17</sup>, Christine Rousset-Jablonski<sup>18,19</sup>, Dominique Sturz<sup>20</sup>, Angela Tincani<sup>14</sup>, Ana Vieira<sup>21</sup>, Susana Capela<sup>12,13</sup>, Dorica Dan<sup>22</sup>, Julie De Backer<sup>23</sup>, Christine de Die-Smulders<sup>24</sup>, Andreas Dufke<sup>25,26</sup>, Estelle Lecointe Artzner<sup>27</sup>, Giuseppe Limongelli<sup>28</sup>, Birgit Lorenz<sup>20</sup>, Wiebke Papenthin<sup>29</sup>, María Jesús Pascau<sup>30</sup>, Johanna Raidt<sup>31</sup>, Isabelle Ray-Coquard<sup>32</sup>, Rachel Rimmer<sup>33</sup>, Claas Röhl<sup>34</sup>, Holm Schneider<sup>35</sup>, Tet Yap<sup>36</sup>, Rosaria Talarico<sup>1</sup> and Marta Mosca<sup>1\*</sup>

#### Abstract

**Background** Rare and complex diseases can have a significant impact on family life, and managing the reproductive aspects of patients of childbearing age with rare diseases is often difficult and complex.

A European Reference Network (ERN) Transversal Working Group (WG) on Pregnancy and Family Planning was created to join forces to promote and address issues on these topics in rare and low-prevalence diseases.

**Objective** To outline the challenges and the good practices related to pregnancy and family planning in rare and complex diseases for healthcare professionals (HCPs).

**Methods** A survey on state of the art and unmet needs was created by a co-design group of both clinicians and patients' representatives from 20 ERNs. The survey was uploaded in English on the online platform "EU Survey" and disseminated by respective ERNs and learned societies. Seven transversal domains were explored in the survey by using closed and open-ended questions: fertility preservation, pre-conceptional counselling, family planning counselling, pre-implantation diagnosis, prenatal diagnosis, pregnancy monitoring and post pregnancy monitoring, lactation monitoring/counselling and newborn management. The questions investigated for each topic were the following: level of importance, activities performed by the centre, clinical challenges, good practice and educational activities.

<sup>†</sup>Dina Zucchi and Diana Marinello have contributed equally.

\*Correspondence: Marta Mosca marta.mosca@unipi.it Full list of author information is available at the end of the article



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**Results** A total of 197 answers were collected from 24 different countries. Unmet needs for HCPs included: the need to improve communication between different HCPs, the lack of predefined organizational pathways, the lack of availability of expert HCPs for some pregnancy-related issues and the need to streamline the care provided among different countries. In addition, the survey underlined the need to improve the educational activities provided to rare disease patients.

**Conclusions** Physicians and patients need to be educated on the emerged unmet needs in order to standardize the information for both HCPs and patients with rare diseases. Educational activities should be considered to help to disseminate information.

Keywords Rare diseases, Pregnancy-related issues, Unmet needs

#### Background

Family planning, pregnancy, childbirth, postnatal period and the management of a newborn child represent complex moments in the life of every woman and family. Managing such a delicate time can be extremely more complex if one lives with a rare disease that can have a significant impact on each of these aspects of family life [1-3].

From the physician's point of view, dealing with and managing the reproductive aspects of patients of childbearing age with rare diseases is often difficult and complex.

One of the main reasons is that there is often a lack of standardised guidelines and protocols for addressing these aspects in patients with rare diseases.

European Reference Networks (ERNs) were launched by the European Commission in 2017 with the aim of tackling low prevalence and rare diseases that require highly specialised treatment and promoting concentration of knowledge and resources through virtual networks involving healthcare providers (HCPs) across the European Union (EU) [4, 5].

An ERN Transversal Working Group (WG) on Pregnancy and Family Planning was created to join forces to promote and address issues on pregnancy and family planning in rare and low-prevalence diseases. The initial actions of the WG included the creation of a survey on state of the art and unmet needs in family planning, reproduction and related themes in order to plan future transversal activities based on the results of the survey.

The aim of this work was to describe the survey results for HCPs to outline the challenges and the good practices related to pregnancy and family planning in rare and complex diseases.

#### Methods

A co-design group of both clinicians and patients' representatives from different ERNs was created and members from different ERNs were engaged, ensuring the transversality of the surveys.

In the WG 20 ERNs were represented: ERN on bone disorder (ERN BOND), ERN on craniofacial anomalies and ear, nose and throat disorders (ERN CRANIO), ERN on endocrine conditions (Endo-ERN), ERN on epilepsies (EpiCARE), ERN on inherited and congenital anomalies (ERNICA), ERN on respiratory diseases (ERN LUNG), ERN on skin disorders (ERN Skin), ERN on adult cancers (EURACAN), ERN on urogenital diseases and conditions (ERN Eurogen), ERN on neuromuscular diseases(ERN EURO-NMD), ERN on eye diseases (ERN EYE), ERN on genetic tumour risk syndromes (ERN GENTURIS), ERN on diseases of the heart (ERN GUARD-Heart), ERN on congenital malformations and rare intellectual disability (ERN ITHACA), ERN on paediatric cancer (ERN PaedCan), ERN on hepatological diseases (ERN RARE-LIVER), ERN on connective tissue and musculoskeletal diseases (ERN ReCONNET), ERN on immunodeficiency, autoinflammatory and autoimmune diseases (ERN RITA), ERN on Transplantation in Children (ERN TRANSPLANT-CHILD) and ERN on Rare Multisystemic Vascular Diseases (VASCERN). Each ERN nominated 1 clinician and 1 European Patient Advocacy Groups (ePAG) Advocate to represent the Network and bring back the discussion to the ERN.

The co-design group discussed the preliminary drafts of the survey, and an updated version was then provided following the comments provided by the co-design group.

The group identified 7 transversal domains to be explored in the surveys:

- Fertility preservation
- Pre-conceptional counselling
- Family Planning counselling
- Pre-implantation diagnosis
- Prenatal diagnosis
- Pregnancy monitoring
- Post pregnancy monitoring, lactation monitoring/ counselling, newborn management

The survey included closed and open-ended questions and mainly explored for each domain: level of importance (response options were: very important, important, moderately important, slightly important and not important), activities performed by the centre, clinical challenges, good practice and educational activities related to the selected topics.

The complete questionnaire, specifically designed for HCPs, is listed in supplementary material S1. For each domain there were a maximum of 9 questions.

The survey was anonymous and was developed in English on the online platform "EU Survey" [6]. HCPs survey was mainly disseminated by respective ERNs and scientific and professional societies, while patient's survey was made available across social media as well as across different patients' associations, also with the support of the panel of patients' representatives.

The survey was launched on 28th February and closed on 31 July 2022.

Since the survey was completely anonymous and no personal information was collected, an approval of the Institutional Board Review was not needed and the participant's consent to the study was obtained by replying to the survey.

The answers were evaluated using descriptive statistics, and the results of a preliminary analysis were shared and discussed with the members of WG after closing the survey.

#### Results

After the launch of the survey, a total of 197 answers from HCPs were collected with different contributions from the ERNs (Table 1).

Respondents were from 24 different countries (Austria, Belgium, Bulgaria, Czechia, Croatia, Cyprus, Denmark, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Luxemburg, Malta, Netherlands, Poland, Portugal, Slovenia, Spain, Sweden), and the majority were from Italy (21.3%). In addition, 8 answered "Other" at question "In which country is your centre based?".

The level of importance provided to each domain by the respondents are detailed in Table 2, and the percentage of the answers "very important/important" and "not important" on the total responses for each ERN are reported in Supplementary Material (Figure S2–S6).

Overall, the topics indicated as important by the highest percentage of respondents were "pre-conceptional counselling" and "pregnancy monitoring" (83.3% and 75.6%, respectively), while the lower percentage of "very important/important" was recorded for lactation monitoring/counselling and pre-implantation diagnosis (47.7% and 50.8%, respectively).

#### Table 1 ERN of the respondents

Name of the ERN selected by the respondents	Ν	%
VASCERN	36	14.94%
ENDO-ERN	33	13.69%
ERN ReCONNET	23	9.54%
EURACAN	23	9.54%
EURO-NMD	22	9.13%
ERN GENTURIS	16	6.64%
ERN PaedCan	14	5.81%
RARE-LIVER	14	5.81%
ERN-EuroBloodNet	11	4.56%
ERN BOND	8	3.32%
GUARD-HEART	7	2.91%
ERNICA	6	2.49%
ERN ITHACA	5	2.07%
ERN RND	5	2.07%
ERN CRANIO	4	1.66%
MetabERN	4	1,66%
TransplantChild	4	1.66%
ERN ERKNET	2	0.83%
ERN RITA	2	0.83%
EUROGEN	1	0.42%
ERN LUNG	1	0.42%

In some cases more than one ERN was selected in the same answer

## Fertility preservation, pre-conceptional counselling and family planning counselling

Fertility preservation was considered very important/ important from 61.2% of the respondents, and not important for 12.7%.

The fragmentation of care in several centers was pointed out, and the challenges which emerged from the open questions were timing (especially in cases in which it was necessary to start therapy quickly) and costs (not always covered by National Health Systems).

Pre-conceptional counselling, defined as a counselling before a planned pregnancy, was the topic with the highest rate of importance, since 83.3% of the respondents indicated it as "very important/important", with 100% as outcome on the same question for 8 ERNs (Fig. 2 in Supplementary Material). For only 4.1% of participants the topic was rated as not important.

Organizational issues were the most highlighted challenges for pre-conceptional counselling, and in particular the fact that specialists such as gynecologists and/or geneticists are not always in the same hospital.

Family planning counselling, defined as discussion of contraception or planning the time of pregnancy attempt also related to disease activity, was rated as very important/important by 71.6%, and not important by 5.6% of

Торіс	Rate of relevance (% of respondents)			
	Very important/important	Moderately important/slightly important	Not important	
Fertility preservation	121 (61.42%)	51 (25.89%)	25 (12.69%)	
Pre-conceptional counselling	164 (83.25%)	25 (12.68%)	8 (4.06%)	
Family planning counselling	141 (71.57%)	45 (22.85%)	11 (5.58%)	
Pre-implantation diagnosis	100 (50.76%)	58 (29.44%)	39 (19.80%)	
Prenatal diagnosis	106 (53.81%)	62 (31.47%)	29 (14.72%)	
Pregnancy monitoring	149 (75.63%)	30 (15.22%)	18 (9.14%)	
Post pregnancy monitoring	132 (67.01%)	43 (21.83%)	22 (11.17%)	
Lactation monitoring/counselling	94 (47.72%)	62 (31.47%)	41 (20.81%)	
New-born management	120 (60.91%)	47 (23.86%)	30 (15.23%)	

**Table 2**Rate of relevance of the explored topics

participants. In the same topic, the answers to open-ended questions underlined the difficulties in communication between different HCPs, when multidisciplinary teams were not available, and the organizational issues, in particular the lack of standardized protocols.

#### Pre-implantation diagnosis, prenatal diagnosis and pregnancy monitoring

Approximately half of the responses (50.8%) rated preimplantation diagnosis as very important/important, while 14.7% rated this topic as not important.

The most common problems described in the openended questions were that not all centres/countries perform these procedures and that costs can be high. In addition, regulations may differ from one country to another, and the indications authorised by ethics committees are not the same in all countries.

Similar rates of importance were recorded for prenatal diagnosis (53.8% very important/important, 19.8% not important). Among the services provided by the centers regarding this domain there were screening for fetal aneuploidy, echocardiographic monitoring for patients SSA/SSB+and genetic counselling. However, in several centers procedures are scarcely available.

Pregnancy monitoring was rated as very important/ important by about 75%, and not important by less than 10% of participants. Not all the centers have multidisciplinary teams, and organizational issues and lack of expertise in some hospitals was described in open-ended questions.

#### Post-pregnancy monitoring, lactation monitoring/ counselling, newborn management and educational activities

Topics regarding the period after the delivery, the puerperium, were important for 47.7% to 67.0% of

participants, with the lower percentage for lactation monitoring/counselling and the highest for post-pregnancy monitoring.

Lack of standardized protocols caused the most common problem related to these topics.

From the survey it emerged that less than 50% of centers provide topic-related educational activities to patients with rare diseases.

Educational activities were provided from a minimum of 29.4% to a maximum of 46.2% of cases on account of the different topics, with the lower percentage for pre-impantation diagnosis and the higher for preconceptional counselling. The most frequent kind of educational activities were leaflets/brochures and/or specific face to face educational sessions. In Table 3 there are detailed for each kind of educational activities the minimum and maximum percentage calculated on the total number of answers and regardless of the ERNs.

#### Discussion

Despite the progress in approach to rare diseases, there are still many unmet needs for rare patients, families and caregivers [7], including the management of pregnancy and family planning.

Table 3 Educational activities

Kind of educational activities provided to patients	% (min to max)
Leaflets/brochure and educational materials	5.92-12.27%
Specific face to face education sessions	16.18-20.30%
On-line educational material	2.55-5.39%
Link to useful website	2.59-6.32%
Contact to patient organisation(s) or patient forum	3.02-8.92%
Other	1.24-6.17%
NA	39.78–65.95%

The aim of this work was to analyse the results of an ERN-wide surveys for HCPs launched among different ERNs to promote and address issues on pregnancy and family planning in rare and low-prevalence diseases.

Overall, the results of the survey pointed out some unmet needs for HCPs, and in particular: the need to improve communications between different HCPs and educational activities provided to rare patients, the lack of predefined organizational pathways and of availability of expert HCPs for some issues and the need to streamline the care provided among different countries. Main unmet needs emerged from the survey are reported in Fig. 1.

The importance of multidisciplinary teams and of collaboration between different HCPs with expertise in pregnancy in rare diseases emerged as a very important aspect for all the explored domains. This is in line with previous results related also to other more common medical conditions, for which multidisciplinary team management is recommended to reduce the occurrence of adverse maternal and fetal outcomes [8]. This aspect is clearly demonstrated by a recent systematic review on patients with Systemic Lupus Erythematosus showing that a multidisciplinary care during pregnancy had a positive impact on pregnancy-related complications, on the disease course, and relieved the patients' psychological impact [9].

This is related to the necessity to give the right information/support to the patients and to improve the awareness on pregnancy-related issues. Starting the journey with an appropriate pre-conceptional counselling could be the best way to achieve this aim, but identify the right timing for this kind of counselling is still challenging.

The results related to almost all domains underlined that there is no standardization in the approach to rare patients, and lack of standardized protocols emerged especially for the topics regarding the period after the delivery. The lack of guidelines could be a possible explanation, and developing organizational pathways may help to homogenize the care provided to rare patients in different clinical settings. With regards to rare and complex rheumatic diseases, the absence of formalized care pathways related to pregnancy and family planning emerged also in a previous work aimed at analyzing the answers of HCPs from European and non-European referral centers [10].

For several domains, and especially for fertility preservation, pre-implantation diagnosis and prenatal diagnosis, common issues are the scarce knowledge on these techniques, the lack of centers/countries which provide them and the high costs, not always covered by the National Health Service. In addition, the presence of religious, cultural, and ethical issues remains a challenge.

To improve knowledge on these topics and to standardize the approach to rare and low-prevalence patients, educational activities should be considered to help to disseminate general information. For instance, developing links to online resources such as leaflets in electronic version, or providing contacts for patient groups will certainly help in addressing many issues that emerged from the survey.

For families dealing with prenatal rare diseases, patient advocacy organizations was reported to be very useful

## CLINICIANS UNMET NEEDS IN PREGNANCY AND FAMILY PLANING IN RARE AND LOW-PREVALENCE DISEASES



Fig. 1 clinicians unmet needs in pregnancy and family planning in rare and low-prevalence diseases. HCPs healthcare professionals

for parents also to help in alleviating some of their stress [11], and in cases of children with rare diseases the importance of specific support for all family members and of HCPs training was previously reported (3). Overall, our study showed transversal needs that can be addressed through a standardized approach from an organizational perspective, and that will then need to be personalized according to disease-specific needs.

We acknowledge that in the survey there was no standardization or guidelines for the HCPs when asked to rate the relevance of a topic in their area; what we are proposing is based on expert opinion and this aspect may represent a weakness in the work. However, we think that HCPs expert opinion can provide a useful basis and a necessary starting point to plan future research and initiatives for rare and low-prevalence diseases.

#### Conclusions

Overall, the results pointed out the need to educate both physicians and patients on the basis of the emerging unmet needs. Online resources can be an excellent educational tool, they may help in disseminating and standardizing educational activities in order to homogenize the information for HCPs and patients. Therefore, initiatives in this direction by scientific societies, ERNs and patient associations should be promoted and encouraged.

We acknowledge that there is heterogeneity in patients, diseases, settings and countries but, to our knowledge, this is the first work that pointed out challenges related to pregnancy and family planning regarding all these rare and complex diseases. Overall, all the information matters to improve the management of these diseases.

#### Abbreviations

ERNs	European reference networks
HCPs	Healthcare providers
EU	European Union
WG	Working group
ERN BOND	European Reference Network on bone disorder
ERN CRANIO	European Reference Network on craniofacial anomalies and ear, nose and throat disorders
Endo-ERN	European Reference Network on endocrine conditions
EpiCARE	European Reference Network on epilepsies
ERNICA	European Reference Network on inherited and
	congenital anomalies
ERN LUNG	European Reference Network on respiratory diseases
ERN Skin	European Reference Network on skin disorders
EURACAN	European Reference Network on adult cancers
ERN Eurogen	European Reference Network on urogenital diseases and conditions
ERN EURO-NMD	European Reference Network on neuromuscular diseases
ERN EYE	European Reference Network on eye diseases
ERN GENTURIS	European Reference Network on genetic tumour risk syndromes
ERN GUARD-Heart	European Reference Network on diseases of the heart

ERN ITHACA	European Reference Network on congenital malformations and rare intellectual disability		
ERN PaedCan	European Reference Network on paediatric cancer		
ERN RARE-LIVER	European Reference Network on hepatological diseases		
ERN ReCONNET	European Reference Network on connective tissue and musculoskeletal diseases		
ERN RITA	European Reference Network on		
	immunodeficiency, autoinflammatory and		
	autoimmune diseases		
RN TRANSPLANT-CHILD	European Reference Network on Transplantation in		
	Children		
VASCERN	European Reference Network on Rare		
	Multisystemic Vascular Diseases		

#### Supplementary Information

The online version contains supplementary material available at https://doi. org/10.1186/s13023-024-03435-z.

Additional file1.	
Additional file2.	
Additional file3.	
Additional file4.	
Additional file5.	
Additional file6.	

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#### Author contributions

DZ and DM contributed to the conception and design of the study, to the acquisition and interpretation of data, and were responsible for writing the manuscript. SA, AB, RB, IB, PB, MCB, MLB, EC, JECF, MF, VI, SL, CN, MO, BP, CRJ, DS, AT, AV, SC, DD, JDB, CDS, AD, ELA, GL, BL, WP, MJP, JR, IRC, RR, CR, HS, TY participated in the execution of the study and data collection. CT and GF were responsible for the analysis of the data and contributed to the drafting of manuscript. RT contributed to all aspects of study design. MM contributed to the conception and design of the study and to the acquisition of data. In addition MM was also responsible for the interpretation of data and contributed to the drafting of the manuscript. All authors were responsible for drafting the article and all the authors have read and approved the final manuscript.

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#### Data availability

The dataset generated and analysed during the current study is not publicly available to keep the answers to open questions confidential but is available from the corresponding author on reasonable request.

#### Declarations

#### Ethics approval and consent to participate

An ethics committee approval was not needed since the survey was completely anonymous and completed by clinicians, and patients were not recruited. The participant's consent to the study was obtained by replying to the survey.

#### **Consent for publication**

Not applicable.

#### **Competing interests**

The authors declare that they have no competing interests.

#### Author details

<sup>1</sup>Rheumatology Unit, Azienda Ospedaliero Universitaria Pisana and Department of Clinical and Experimental Medicine, University of Pisa, ERN ReCONNET, Via Roma 67, Pisa, Italy. <sup>2</sup>Department of Medical Biotechnologies, University of Siena, Siena, Italy. <sup>3</sup>Spanish Association for Antiphospholipid Syndrome (SAF España), ERN ReCONNET, Madrid, Spain. <sup>4</sup>Service de Gynecologie Obstetrigue, Hopital Antoine Béclère, AP-HP, Université Paris Saclay, ERN ERNICA, Clamart, France.<sup>5</sup>ERN GUARD-Heart, Coordinator Center, Amsterdam, The Netherlands. <sup>6</sup>ARVC-Selbsthilfe E.V., Munich, Germany.<sup>7</sup>Germans Trias Hospital, ERN GENTURIS, Badalona, Spain. <sup>8</sup>VASCERN, Coordinator Center, Paris, France. <sup>9</sup>Cutis Laxa Internationale, ERN-Skin, Bons-en-Chablais, France.<sup>10</sup>Università Vita-Salute San Raffaele, ERN BOND, Milan, Italy. <sup>11</sup>SAMS Association, ERN GUARD-Heart, Philadelphia, USA. <sup>12</sup>Serviço de Reumatologia Centro Hospitalar Universitário Lisboa Norte, ERN ReCONNET, Lisbon, Portugal. <sup>13</sup>Instituto de Medicina Molecular, Faculdade de Medicina, Universidade de Lisboa, Centro Académico de Medicina de Lisboa, Lisbon, Portugal.<sup>14</sup>Department of Clinical and Experimental Sciences, Rheumatology and Clinical Immunology, University of Brescia, ASST Spedali Civili of Brescia, ERN ReCONNET, Brescia, Italy. <sup>15</sup>Department of Pediatrics, UMHAT "St. Marina", Endo-ERN, Varna, Bulgaria. <sup>16</sup>ERN Euro-NMD, Coordinator Center, Paris, France. <sup>17</sup>CDH UK-The Congenital Diaphragmatic Hernia Charity, ERNICA, King's Lynn, UK.<sup>18</sup>Department of Surgery, Leon Berard Cancer Center, Lyon, France. <sup>19</sup>INSERM U1290 RESHAPE (RESearch in HealthcAre PErformance), EURACAN, Lyon, France. <sup>20</sup>ERN-Eye, Coordinator Center, Strasbourg, France.<sup>21</sup>Liga Portuguesa Contra as Doenças Reumáticas, Núcleo de Sjögren, ERN ReCONNET, Lisbon, Portugal. <sup>22</sup>ERN ITHACA, Coordinator Center, Paris, France. <sup>23</sup>Department of Cardiology and Center for Medical Genetics, Ghent University Hospital Belgium, VASCERN, Ghent, Belgium. <sup>24</sup>GROW, School for Oncology and Reproduction, Maastricht University, ERN NMD, Maastricht, Netherlands. <sup>25</sup>Institute of Medical Genetics and Applied Genomics, University of Tübingen, Tübingen, Germany.<sup>26</sup>Centre for Rare Diseases, University of Tübingen, ERN ITHACA, Tübingen, Germany.<sup>27</sup>Sarcoma Patients Advocacy Global Network (SPAGN), ERN EURACAN, Berlin, Germany. <sup>28</sup>Inherited and Heart Disease Unit, Monaldi Hospital, AO Colli-University of Campania "Luigi Vanvitelli", ERN GUARD-Heart, Naples, Italy.<sup>29</sup>ERN RARE-LIVER, Wilson Working-Group, Pittsburgh, USA. <sup>30</sup>Paediatric Transplant Unit, La Paz University Hospital, ERN Transplantchild, Madrid, Spain. <sup>31</sup>Department of General Pediatrics, University Hospital Muenster, ERN LUNG, Muenster, Germany. <sup>32</sup>Centre Leon Bérard, Université Claude Bernard Lyon Est, ERN EURACAN, Lyon, France. <sup>33</sup>ERN RITA, Coordinator Center, Utrecht, The Netherlands. <sup>34</sup>NF Kinder, Vienna, Austria. <sup>35</sup>University Hospital Erlangen, ERN Skin, Erlangen, Germany. <sup>36</sup>Guy's and St Thomas' NHS Trust, ERN eUROGEN, London, UK.

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