RARE 2030

D2.4 European "Consensus Conference" Report

January 2021

Report Information

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Project Information

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European "Consensus Conference" Report

Introduction

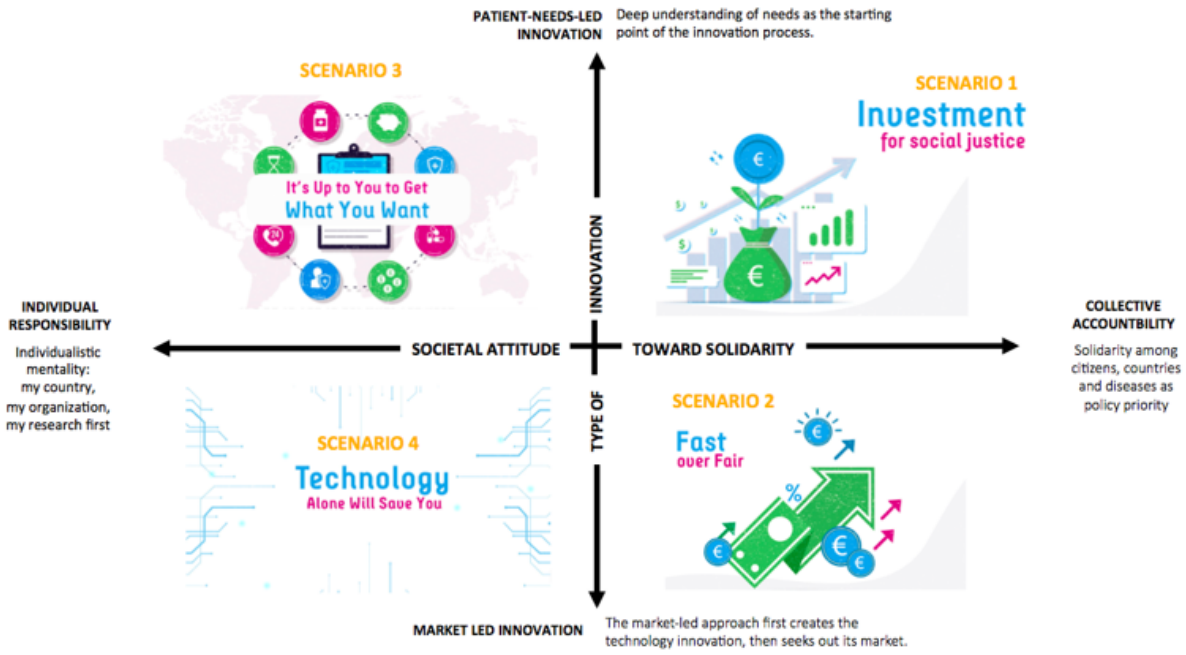
As part of the Rare2030 Foresight project and in order to consolidate the Rare2030 scenarios, we sought to engage young citizens interested in the field of rare diseases in the policy debate through a series of online meetings, conferences and capacity building opportunities to prepare them to provide their opinion on the future of rare disease policy.

Following a series of workshops and assignments spread from March to July, a two-day online meeting was organised on July 7th and 8th with 10 experts and 23 young citizens – bridging the gap between the next generation public and experts to contribute to and validate a series of future policy scenarios at the 2030 horizon of rare disease policy. A final online event, the Young Citizens Fall Debate, was also organised on October 28th, which gave Young Citizens the opportunity to present their consolidated recommendations to a jury.

On the occasion of the conference, the participants focused on 8 topics of discussion identified in earlier stages of the Rare2030 project:

1. Political and strategic frameworks relevant to rare diseases
2. Data collection and utilisation
3. Availability and accessibility of orphan medicinal products (OMPs) and medical devices
4. Basic, clinical, translational and social research
5. Diagnostics
6. Integrated, social and holistic care for people living with a rare disease
7. Rare disease patient partnership
8. Access to healthcare

The participants discussed and debated over these topics through the lens of the Rare2030 policy scenarios, with a particular emphasis on scenario 1 ('Investment for Social Justice') and scenario 2 ('Fast over Fair') as these were the scenarios the rare disease community voted on as being the most preferred ('Investment for Social Justice') and the most likely if we continue as we are now ('Fast over Fair').



Participants

Application and selection criteria

We sought to gather the opinions of youth from across as many European countries as possible, spanning sectors such as medicine, nursing, social work, human rights, policy, public health, etc. as well as siblings of patients and patients themselves.

Following an open call to apply, young citizens were recruited from EURORDIS' network of advocate members, recommendations from universities and platforms, project partners, and contacts from different national and European bodies. The criteria taken into consideration for the selection of participants were as follows:

- Aged between 18 and 30 years old
- From a European country
- With a strong interest in and motivated by the rare disease cause
- Being fluent in English
- With one of the following fields of expertise and/or background: patient representatives, advocate members, siblings of people affected with rare diseases, students, doctors, health professionals, health policy, public health, health economics, human rights or rare disease patient advocates themselves.

To apply as a potential young citizen, candidates were requested to fill in an online form (see Annex 1)

Selection of participants

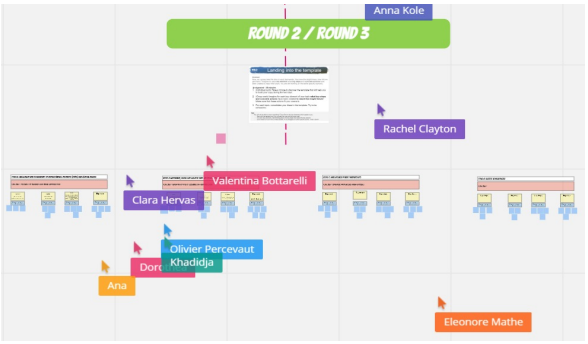
Participants were selected based on the abovementioned criteria and, while most of them fulfilled these criteria, two candidates living outside of Europe were eventually not selected. In addition to that, with regards to the enthusiasm displayed, we expanded the age limit to 32 years old, allowing us to select 28 people for the conference – which includes two people being 31 years old (see Annex 2 for full profiles).

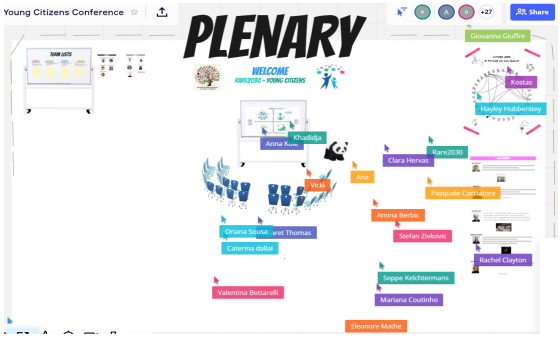
However, with the outbreak of COVID-19 having forced us to move the Young Citizens Conference online and to divide the programme in two days, five initially selected candidates were no longer available to take part into the conference because of clashing work and/or personal commitments.

Structure of the Conference

We sought to structure the up-stream engagement of young citizens in discussions through a modified "citizens conference". Traditionally, citizen conferences occur between citizens and experts over a series of several weekends to build the capacity of citizens to formulate and structure their arguments, and ultimately come up with a point of view from this public. Rather than following a strict "citizens conference" methodology, we sought to invite this panel of young citizens to a series of online meetings, conferences and capacity building opportunities to prepare them to provide their opinion on the future of rare disease policy.

Throughout these workshops and events, we used an online collaborative whiteboard platform, Miro, which allowed participants to discuss and share opinions as if they were in the same room. The principle of this program is to display an infinite interactive whiteboard to facilitate brainstorming and presentation of discussions outcomes online.





Capacity building workshops and trainings

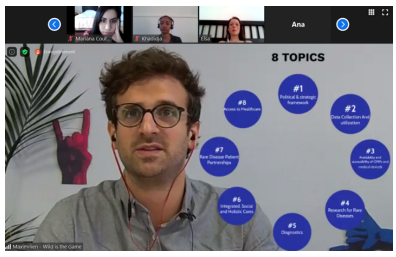
1. Preparatory webinars

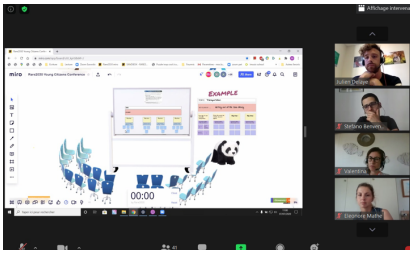
Following our modified approach of citizens conferences, we organised three webinars in the course of March – May to prepare young citizens to take part into the conference. The goals of these webinars were to introduce participants to the Rare 2030 project and to highlight the issues around rare disease policies. One of these workshops was specifically designed to teach young citizens to effectively use Miro.

- **Webinar 1**: Get ready, get set... for the Rare2030 Young Citizens Conference - Rare2030 Project and advocacy: an introduction (see presentation in Annex)
  This webinar introduced Rare2030 Young Citizens to the project, its objectives, methodology and the important role they play in the process

- **Webinar 2**: Rare diseases 101: context and challenges
  This webinar went into greater detail of the foresight reflections having taken place to date, highlighting current challenges for people living with rare disease and for those working in industries to help them

- **Webinar 3**: Whistle while we Work: introduction to Miro
  This webinar has allowed young citizens to get familiar with the programme Miro through small online exercises





2. European Conference for Rare Diseases and Orphan Drugs (ECRD)

Young Citizens were invited to take part into the ECRD that took place online on May 14th and 15th. Rare 2030 being at the center of this edition of the ECRD, young citizens were requested to attend 4 sessions in addition to the opening and closing plenary sessions. On this occasion, they were presented with a video that illustrates the four different possible scenarios (‘*Investment for social justice*’, ‘*Fast over fair*’, ‘*It’s up to you to get what you want*’, and ‘*Technology alone will save you*’) defined in earlier stage of the Rare 2030 project. The overall structure of the conference was divided into six themes, which were subdivided into four specific sessions and gave young citizens the opportunity to focus on their topic of choice. Following the ECRD, young citizens were asked to provide us with a feedback that included:

- Their opinion on the Rare 2030 scenarios
  - Which one they like the best?
  - Which one they like the least?
  - Which one is, according to them, most likely to take place?

- A summary of two sessions they attended
- A brief text on the policy options they would recommend with their current knowledge
- A general feedback on their experience at the ECRD

3. The Young Citizens Conference

The Young Citizens Conference took place online on July 7th and 8th. Initially scheduled to be a full day of workshops and presentations in Barcelona, we moved the conference online due the COVID-19 pandemic and divided the initial one-day event in two consecutive mornings.

Prior to the conference, young citizens were divided into four groups and assigned specific topics. The groups and topics are displayed below:

- **Group A**: *Political and strategic framework for RDs/Diagnostics*
  - Julien Delaye, Public Affairs Assistant, Belgium
  - Danielle Drachmann, Executive Director/Patient advocate, Denmark
  - Pasquale Cacciatore, Medical Doctor, Italy
  - Fanni-Laura Mäntylä, Patient Advocate/Health Activist, Finland
  - Alessandra Blonda, PhD Student, Belgium
  - Seppe Kelchtermans, Policy Advisor, Belgium

- **Group B**: *Availability and accessibility of OMPs/integrated social and holistic care*
  - Rachel Clayton, French/International Relations Student, UK
  - Khadidja Abdahllah, PhD Researcher, Belgium
  - Amina Berbic, Project and Financial Manager, Bosnia and Herzegovina
  - Ana Begic, Global Regulatory Affairs Officer, Croatia
  - Eleonore Mathe, Pharmacy Doctor, UK
  - Dorothea Dalig, Public Affairs Consultant, Belgium

- **Group C**: *RD patient partnerships/Access to healthcare*
  - Marie Barth, PhD Biochemistry Student, Germany
  - Stefan Zivkovic, Office Assistant, serbia
  - Mariana Coutinho, Clinical Psychology Researcher, portugal
  - Rita Francisco, PhD Molecular Genetics Student, Portugal
  - Thomas Villaret, Law Student
  - Elsa Coulomb, Pharmacist, France

- **Group D**: *Diagnostics/integrated, social and holistic care*
  - Caterina Dallai, Psychology Student, Italy
  - Hayley Hubberstey, Health Economist, UK
  - Oriana Sousa, Advisory Board Member, Portugal
  - Sara Cavalleri, Medical Student, Italy
  - Aneta Morkovska, Doctor, Czechia

In addition, we invited experts in the field of rare diseases to be part of a jury, whose responsibilities were both to assist young citizens in the formulation of Young Citizens' ideas and policy recommendations, as well as to provide them with a feedback following the presentation of their work. Composition of the jury is displayed below:

- Kostas Aligiannis, Public Affairs Manager, EURORDIS
- Charlotte Rodwell, Partnerships, Business Development and Strategic Communications Officer, Orphanet
- Victoria Hedley, Rare Disease Policy Manager, Newcastle University
- Simone Boselli, Public Affairs Director, EURORDIS
- Giovanna Giuffrè, project Manager, Isinnova
- Ines Hernando, ERN and Healthcare Director, EURORDIS
- Anna Kole, Rare 2030 Project Lead, EURORDIS
- Valentina Bottarelli, Public Affairs Director, EURORDIS
- Clara Hervas, public Affairs Manager, EURORDIS
- Stefano Benvenuti, Global Partnership Manager, Fondazione Telethon

The two days were structured as a series of small workshops designed to help young citizens identify major issues in their assigned topics, brainstorm on how these challenges could be addressed and eventually formulate policy recommendations to improve the life of people living with a rare disease in the next ten years. The goal of these successive workshops was to enhance a truly iterative and interactive process, with initially raw ideas maturing with each new step. Following this logic, steps of the process are described below:

- *Step 1*: Identifying major issues within the different topics and ranking them according to young citizens' perception of importance
- *Step 2*: Identifying, for each selected challenges, what major obstacles one would encounter when attempting to solve these issues, as well as which stakeholders would have a role to play to do so
- *Step 3*: Defining the steps that should be taken to address specific issues
- *Step 4*: Refining these steps and proposing actual policy recommendations
- *Step 5*: Presenting these recommendations to a panel of experts

4. The Young Citizens Fall Debate

On October 28th, young citizens and a jury constituted of experts in the field of rare diseases gathered once more online on the occasion of the Young Citizens Fall Debate. Prior to the event, young citizens were asked to refine their work based on the feedback they received from the Young Citizens Conference jury and to prepare group presentations that focus on their most important recommendations.

Groups remained the same, with two young citizens presenting per group (see annex 3 for the group presentations). However, to allow for a different feedback from the Young Citizens Conference, we invited a different panel of expert to join in the quality of jury:

- Yann le Cam, Chief Executive Officer, EURORDIS
- Kate Bushby, Clinical Academic
- Inês Alves, Patient representative and ERN-BOND member
- Rebecca Stewart, Co-Founder and Chief Executive Director, Rare Revolution Magazine
- Allan Sam, Parliamentary Advisor, European Parliament

5. The Young Citizens Open Letter

Based on the outcomes of both the Young Citizens Conference and Fall Debate, we wrote an open letter addressed to policy makers in the name of Young Citizens. This letter is intended to highlight what young citizens – in their quality of current and future patients/patient advocates, doctors/nurses/healthcare providers, and policy makers – hope and expect from the future to improve the lives of people living with a rare disease in Europe. The Open Letter can be found in Annex 4.

Results

Young Citizens' views on Rare 2030 scenarios

Young Citizens watched the scenario video presented on the occasion of the ECRD and unanimously considered scenario 1 (*Investment for Social Justice*) as their most preferred scenario. Scenarios 4 (*Technology Alone Will Save You*) and 2 (*Fast Over Fair*) were considered as the scenarios that would be the least beneficial to people living with a rare disease. Lastly, scenario 2 was also said to be the most likely to happen in the future if no major policy or effort is implemented, therefore being identified as a potential threat to the rare disease community in the next ten years.

Young Citizens' policy recommendations

Following the Young Citizens Conference and Fall Debate, young citizens propose the following policy recommendations.

Political and Strategic Frameworks relevant to Rare Diseases

- ***Establish a European Rare Disease agency***

  Young citizens recommend gathering expertise, means and resources into one single European agency that would be managing and responsible for all aspects of rare diseases across Europe, encompassing funding, social support, market access, data management, disease/patient registries to allow for a continuum in these activities and ensure collaboration, interoperability and use of best practices.

- ***Develop a more cohesive and more transparent reimbursement process, and fostering European collaboration***

  Young citizens believe individuals do not have a clear understanding of reimbursement procedures across Europe and recommend further steps in the alignment/clarification of reimbursement procedures. While these procedures vary from one country to another and remain the responsibility of member states, they however also recommend developing a European common system that would pay healthcare professionals for the time they spend with foreign patients and simplifying the related reimbursement process.

- ***Establish a European reward system for treatment/care abroad***

  To foster cross-boarder healthcare, young citizens recommend establishing a system of reward to countries when they care for foreign patients, while ensuring transparency and legitimacy. They

believe developing a common European funding scheme for rare disease treatment at the EU level could bridge the gap between patients' needs and national resources to ensure care for all.

- ***Improve cross-country collaboration***
  In line with recent projects and political efforts, young citizens recommend further sharing of best practice, collaboration on topics such as reimbursement procedures or access to care abroad.

- ***Develop more company-oriented incentives specifically for diagnosis***
  Furthermore, more intervention from the EU to foster collaboration on diagnosis (structure, funds) has also been recommended.

- ***Address discrimination through political framework and efforts***
  With rare diseases being often misunderstood and stigmatized, young citizens recommend the followings on the topic of discrimination:
  a) Develop training on discrimination and how to deal with it in different settings (hospitals, social sector, etc)
  b) Launch an awareness campaign to reduce stigmatisation
  c) Create laws and policies targeting discrimination and stigmatisation, thus making states more accountable for discrimination issues.

- ***Address mental health issues and social care challenges***
  As being diagnosed with a rare disease is challenging in many respects, young citizens support the setting up of mental health support upon diagnosis. Going further, they also recommend providing increased funding to hospitals with regards to social care rather than solely on healthcare per se.

- ***Develop a legal framework for data ownership and protection***
  Data being at the center of research, young citizens recommend developing a framework that takes account of new technologies and what their use may entail.

- ***Improve articulation between public and private sectors***
  Echoing other recommendations, young citizens suggest clarifying and developing the roles of public and private sectors to guarantee optimal collaboration, which would thus positively impact access to care in general.

- ***Develop mixed funds scheme and practices for patients who need treatment from abroad***
  Young citizens believe that such practices should be encouraged and financed by governments.

- ***Foster joint-HTA assessment***
  Recent developments and projects on joint-HTA assessments have proven to be highly beneficial to the rare disease community. However, as these project often only concern a relatively small number of countries, young citizens recommend fostering such effort and go beyond the scope of a few countries, hence the importance of European collaboration to guarantee that the most cost-effective, innovative and safest technologies are available in Europe,

Data Collection and utilization

- ***Improve cross-country collaboration***
  Despite many advances in the last decade the young citizens recommend further sharing of best practice and collaboration across countries and sectors to improve a continually siloed data landscape.

- ***Develop a legal framework for data ownership and protection***
  In this new technological era with the development of big data and the continuous sophistication of information and communication technologies, and the increasingly recognized potential of data in health and well-being solutions, young citizens recommend developing a framework that takes account of new technologies and applications.

- ***Establish patient-centricity as the standard model of research and development in Europe***
  With limited knowledge on rare diseases and limited resources as compared to other disease areas, the young citizens recommend that putting patients at the center of research will ensure that existing data and knowledge is best used.

Availability and Accessibility of Orphan medicinal products and Medical Devices

- ***Develop a more cohesive and more transparent reimbursement process, and fostering European collaboration***
  Young citizens believe individuals do not have a clear understanding of reimbursement procedures across Europe and recommend further steps in the alignment/clarification of reimbursement procedures. While these procedures vary from one country to another and remain the responsibility of member states, they however also recommend developing a European common system that would pay healthcare professionals for the time they spend with foreign patients and simplifying the related reimbursement process.

- ***Establish a European reward system for treatment/care abroad***
  To foster cross-boarder healthcare, young citizens recommend establishing a system of reward to countries when they care for foreign patients, while ensuring transparency and legitimacy. They believe developing a common European funding scheme for rare disease treatment at the EU level could bridge the gap between patients' needs and national resources to ensure care for all.

- ***Improve cross-country collaboration***
  In line with recent projects and political efforts, young citizens recommend further sharing of best practice, collaboration on topics such as reimbursement procedures or access to care abroad.

- ***Facilitate authorization process and improving social support to patients while seeking care abroad.***

- ***Foster joint-HTA assessment***
  Recent developments and projects on joint-HTA assessments have proven to be highly beneficial to the rare disease community. However, as these project often only concern a relatively small number of countries, young citizens recommend fostering such effort and go beyond the scope of a few countries, hence the importance of European collaboration to guarantee that the most cost-effective, innovative and safest technologies are available in Europe.

Diagnostics

- ***Supporting those with "undiagnosed" rare diseases***
  More multistakeholder work is required to achieve solutions for "hard to diagnose rare diseases".

- ***Improve existing newborn screening programmes***
  The young citizens recommend that more research should focus on comparing different national newborn screening approaches to highlight the benefit of expanding the scope of rare diseases included in national screening programmes.

- ***Increased investments in diagnostic solutions***
  Young Citizens recognise that diagnosis is the first moment when people living with a rare disease are recognised and registered. As such they recommend that European level patient registries are linked to this source of diagnosis information. Such efforts would require action and investments both at the European (harmonising and linking data) and national levels (collecting and sharing data). Currently private investments for improving the life of people with rare diseases are focused on treatments. Young Citizens call for more incentives for private investments in diagnostic innovations. These incentives should go beyond current sponsors of rare disease treatments.

- ***Employ telemedicine and remote care to support improved diagnosis***
  The Young Citizens consider that the COVID-19 crisis has demonstrated the enormous potential for telemedicine in delivering healthcare. Fostering efforts and further developing telemedicine and remote care practices would allow patients to have appointments and tests for diagnostic purposes without having to travel unnecessarily.

- ***Solidify pathways from first symptoms to care***
  Young citizens believe that ERNs should play a central role with hospitals for the development of protocols and 'flowchart' of symptoms that would facilitate diagnosis and increase awareness on rare disease diagnosis.

Integrated, Social and Holistic Care for People With Rare Diseases

- ***Reframe what it means to be a person living with a rare disease***
  In line with the theme of Rare Disease Day 2020, young citizens recommend reframing what it means to live with a rare disease, taking into account domains - beyond clinical care and treatments - such as holistic care and the daily needs that are currently either not clearly

understood or addressed in current policies (e.g. mental health, sexual health or idiopathic disease or symptoms).

- ***Enhance education around rare diseases and their challenges***
  Young citizens believe education and awareness on disability and the holistic need of people living with rare diseases is not sufficiently emphasised in society. They recommend putting additional efforts and resources to ensure a curriculum in schools that addresses these topics at a younger age. Greater awareness on rights to cross-border and holistic care - rights that are still often misunderstood or unknown by healthcare professionals, hampering potential benefits - is required for an effective implementation in the upcoming decades -. With rare diseases being often misunderstood and stigmatized, young citizens recommend more awareness raising campaigns to reduce the stigmatisation of people living with a rare disease and their families in society. They also recommend increased development of training on discrimination and better implementation of existing trainings in all appropriate settings (hospitals, social sector, schools, etc.).

- ***Improve assessment methods***
  The importance of research to demonstrate the value of and improve holistic care should be acknowledged and increasingly supported. People living with a rare disease should be involved in designing social research to best address priorities. Tools to collect patient reported outcomes and experience (PROMs and PREMs) should be developed to allow reporting of outcomes and unmet needs. Expert advocates should be part of research and of hospital structures in order to more efficiently and effectively guarantee the holistic nature and integration of services.

- ***Better address psychological and social challenges for people living with a rare disease in society***
  Funding to integrate a holistic approach in hospitals and other healthcare settings should be increased. Mental health services should be well integrated, particularly alongside diagnostic services. This should be fostered both by legislation and policies in the private and public sectors. Legislations and policies addressing the challenges people and families face in living with a rare disease should be improved particularly in targeting discrimination and stigmatisation and making countries more accountable in their implementation.

<u>Rare Disease Patient partnerships</u>

- ***Promote partnership between actors***
  More specifically, young citizens recommend increasing partnership between patient organisations and academic institutions through programs to discover new tests and technologies, while lobbying for them at the same time (mostly for diseases for which there exist no diagnosis options).

- ***Stimulate networking to increase awareness***
  Because many doctors do not know enough about rare diseases and are therefore unable to accurately and effectively diagnose most of them, Young Citizens recommend increasing the networks that already exist and define them as key centers. These key centers could coordinate care in cases where diagnosis is too complex and could also establish a list of key stakeholders

around Europe to facilitate coordination and expertise. Furthermore, organising an annual meeting gathering key actors and groups to facilitate coordination and expertise has also been recommended.

- ***Enhance education at younger age***
  Young citizens believe education on general health is not sufficiently emphasized. Hence, they recommend putting the stress on improving education at younger age and further acknowledging the importance of holistic research and care, as well as the benefits this could have on people living with a rare disease.

- ***Improve needs assessment***
  Young citizens recommend including the opinion of people living with a rare disease in identifying research priorities to ensure they align. Young citizens suggest the development of tools or platforms to fill gaps in current research, care and treatment availability and access. Specifically, expert patients should be part of research and of hospital structures in order to support that research and care targets directly address patient needs and help define the best processes in doing so.

- ***Develop training and educational program for all stakeholders***
  Specifically, young citizens point out to cultural differences and barriers that should be addressed in such trainings.

- ***Create, validate and promote the role of the 'expert patient'***
  Young citizens recommend that the "expert patient" be an official role and job position across sectors contributing to rare diseases, which would comply with ethical and legal guidelines to ensure no conflict of interest and be formally acknowledged and paid.

- ***Establish bi-directional specialised department for partnership in both companies and patient organisations***
  Alongside the recommendation of promoting the role of the "expert patient", Young Citizens recommend that bi-directional units/departments specialised in partnership should exist in healthcare industry companies and patient organisations.

- ***Improve articulation between public and private sectors***
  Echoing other recommendations, young citizens suggest further encouraging collaboration between public and private sectors including civil society and patients to guarantee optimal collaboration, ultimately leading to better quality policies, programmes and services.

Access to Healthcare

- ***Clarify and reframe the role of ERNs***
  The young citizens recommend clarifying and better communicating how an individual person living with a rare disease could adequately benefit from these networks as they believe patients

and physicians do not currently have a clear understanding of ERNs' benefits and structure nor do they understand how they can interact with them.

- ***Establish a European reward system for care and treatment abroad***
  To foster cross-border healthcare, young citizens recommend facilitating a smoother authorization process for treatment abroad. They suggest establishing a system of reward for countries when they care for foreign patients, while ensuring transparency and legitimacy. They believe developing a common European funding scheme for rare disease treatment at the EU level could bridge the gap between patients' needs and national resources to ensure care for all. This mechanism could also improve social support to patients while seeking care abroad.

- ***Improve cross-country collaboration***
  In line with recent projects and political efforts, young citizens recommend further sharing of best practice, collaboration on topics such as reimbursement procedures or access to care abroad.

- ***Improve telemedicine practices***
  As the COVID-19 crisis has demonstrated, there is an enormous potential for telemedicine to be used in healthcare. Fostering efforts and further developing telemedicine practices would allow patients to have remote appointments without having to travel and, in a second time if adequately developed, benefit from telemedicine for care.

Conclusion

Young citizens clearly expressed their preference for scenario 1, "*Investment For Social Justice*", particularly with regards to aspects of solidarity across countries and disease areas. However, they also believe that, should no further major policies be implemented in the upcoming years, we would be likely to fall into scenario 2, "*Fast Over Fair*". In both cases, their opinion echoes other stakeholders' point of view with regards to the Rare 2030 scenarios.

As shown earlier, young citizens have proposed different policy recommendations, addressing the Rare 2030 topics, to improve the lives of people living with a rare disease in the next ten years. When compared to other stakeholders' recommendations, the ideas and opinions stemming from the youth seem to be in line with other cohorts' findings. Collaboration across countries and fields of expertise appears as the most prominent condition for policy recommendations to be successful and useful to the rare disease community. Despite trends in nationalism and threat to European solidarity, young advocates give us faith in their common vision for increased collaboration across Europe. The role and use of ERNs have come frequently come up in discussions, described as unclear by most young citizens and the broad topic of collaboration, spanning across the 8 themes of Rare 2030, was also repeatedly addressed and particularly emphasised during presentations and debates, which brought both young citizens and jury members to agree that collaboration is a vital component of future policy developments.

Young citizens also put the stress on holistic care, particularly when it comes to mental health or education of social and health care providers. Regarding mental health, young citizens specifically point the time of diagnosis as a major opportunity for improvements in terms of psychological support, while

education should be fostered mostly at the (health) professional level through trainings. Lastly, they recommend new assessments methods that take account of holistic care alongside treatments and therefore support the development of new tools and means to evaluate a situation beyond physiological terms. Once again, these findings are in line with other stakeholders' recommendations and previous findings.

Hand in hand, the recommendations of young citizens tend to support the recommendations of the Rare 2030 project. We have observed convergence of findings and recommendations between stakeholders, which in turn reinforces the need to address the gaps that exist in the current policy framework. More importantly, the fact that the next generation of patients/patient advocates, health and social care providers, and policy makers seem to agree on the next priorities is an encouraging sign that more efforts could be achieved on commonly agreed topics.

Lastly, it is important to highlight the secondary benefits of the Young Citizens Conference. While it has allowed us to include youth into the project and come up with a set of recommendations supporting the work of Rare 2030, it has also led to identify highly motivated members of the next generation of advocates from different background and expertise. The dynamic of the group we have observed throughout these past few months clearly demonstrates that policy changes need to happen at various levels and that young individuals not only agree on policy priorities but also show willingness to be part of process that will build a better future for people living with a rare disease.

Annex

Annex 1: Invitation to Young Citizens

Rare 2030 YOUNG CITIZEN CONFERENCE

Application Form

I. START

What is the Rare2030 Young Citizen Conference?

As part of the Rare2030 project, we seek to engage young citizens interested in the field of rare diseases on the future policy.

The Conference will take place in Barcelona on 8 July 2020 in EURORDIS-Rare Diseases Europe premises.

With the help of an expert moderator in citizen engagement, the conference will bring together young citizens, experts and other stakeholders to contribute to validate a series of potential future policy scenarios at the 2030 horizon of rare disease policy on the specific topics such as:

- Political & strategic frameworks relevant to rare
- diseases Data collection and utilisation
- Availability and accessibility of orphan medicinal products and
- medical devices Basic, clinical and translational research
- Diagnostics
- Social integration of rare disease patients and
- holistic care Patient engagement
- Accessing healthcare

A series of events preceding the Young Citizen Conference will be organised:

- 2 online webinars on 12 and 26 March
- European Conference on Rare Diseases and Orphan Drugs (ECRD) on 15-16 May in Stockholm

Participants will be expected to attend all the events and participate in the different planned activities. The Young Citizen Conference is open to approximately 30 young citizens in Europe.

What can this opportunity offer you?

- Advocacy and policy making experience
- Working in multidisciplinary environment

- Make your voice matter
- Full expenses covered (travel and accommodation covered in accordance with EURORDIS' reimbursement policy)

How to apply?

• Fill in this online application by **1 March 2020**
• First come, first served

What are the selection criteria?

• The candidate has to be from a European country (priority given to EU countries)
• Related fields of expertise: patient representatives, advocate members, siblings of people affected with rare diseases, students, doctors, health professionals, health policy, public health, health economics, human rights.
• Motivated by rare disease cause
• Interested in advocacy
• Must be able to participate in full process: 2 online webinars on 12 and 26 March, European Conference on Rare Diseases and Orphan Drugs (ECRD) on 15-16 May in Stockholm, Rare2030 Young Citizen Conference on 8 July in Barcelona
• English speaking
• Age requirements: 18-30

II. PERSONAL INFORMATION

* First name

* Last name

* Name of your organisation / group / university

* Your position / role in the organisation / group / university

* Short description of your organisation

* Email address

* Country of residence

* What is your age?

at most 1 choice(s)

- [ ] 18-21
- [ ] 22-25
- [ ] 26-30

* Phone number

III. MOTIVATION

* What is your motivation to participate in the Conference? Why should you be chosen?

600 character(s) maximum

IV. EXPERIENCE

* Please describe any relevant experience you may have.

600 character(s) maximum

Please upload any additional file that you may think can support your application (CV, publications, web articles, YouTube videos, Facebook posts, exhibitions, seminars, etc.)

Please upload your file

The maximum file size is 1 MB

In case there are more than one file please name the files with your surname when uploading them.

V. APPLICATION COMPLETE

Thank you for completing your application to attend the Rare2030 Young Citizen Conference. This application will be reviewed and you should receive an answer within a few weeks. Please be aware that your application may be unsuccessful and you should await the receipt of confirmation before making any travelling arrangements.

For general questions regarding the Rare2030 project, please contact Anna Kole, Rare2030 Project Lead at anna.kole@eurordis.org.

For any other questions (logistics, reimbursement, organisation), please contact rare2030@eurordis.org.

Annex 2: Profiles of Young Citizens

AMINA BERBIĆ, project and financial manager, Bosia and Herzegovina

"My name is Amina Berbić. I come from Bosnia and Herzegovina. I am 24. I was born with VSD and ASD. At age of 17, I was diagnosed with pulmonary hypertension due to late repaired of large VSD. As young teenager, I straggled with my disease because I was not "normal" as other teenagers. I tried to avoid talking about my symptoms and disease because I had feeling that I am not sick if I am not talking about it. I have a rare disease thus it was difficult to find other young patients to relate with them. When I managed to find two beautiful girls which gave me strength to fight for my dreams and inspire other patients to do same thing. In that moment I knew that I will do everything I want, not letting my disease define me. What I wanted to say is that I don't want my disease and struggle to make me a victim. I want my battle to make me someone else's hero. Most of my goals come true; I finished my bachelor degree in international finance and master degree in business administration. Also, I learned different art techniques such as oil painting, decoupage technique painting, silk painting, ebru, calligraphy, aluminium foil painting and I started to grow and foster organic food. I keep going and discovering new things. I support and inspire young patients to do same thing within PH Serbia organization, to follow their dreams and achieve their goals. I am working as project manager and financial manager at local women organization called Mosaic of culture and traditions, where I have implemented more than 10 projects and still working on expanding my project portfolio. I want to maximize my leadership potentials in order to success in my goal to create organization for young patients with pulmonary hypertension in B&H. I would like to learn more about ways to help patients to make decisions about their health care and get financial, legal and social support."

ANA BEGIC, senior regulatory affairs officer, UK

"My name is Ana Begic. I've earned my Master of Pharmacy degree from Faculty of Pharmacy and Biochemistry, University of Zagreb (Croatia) in 2013. Following completion of MPharm I enrolled in Postgraduate specialist study Molecular diagnostics to widen my knowledge in pharmacogenomics, personalised medicine and the way human genomics is reshaping our approach to therapy and diagnosis. I am also finalising a Postgraduate specialist study Leadership & Management of Health Services (Faculty of Medicine, University of Zagreb) which was primarily focused on scientific knowledge in the field of health systems and services and their application in clinical management of patient and disease management in accordance with the principles of evidence-based medicine. After working as a pharmacist in a community pharmacy for 3 years, I have joined European Medicines Agency in London as a trainee and a

Procedure Manager in Human Medicines Evaluation, Procedure Management Department. I currently work as a Global Regulatory Affairs Officer in a Contract Research Organization committed to identifying, understanding and addressing unmet public health needs. I want to use my knowledge, enthusiasm and passion to help rare diseases patients together with people who have the power to make a difference."

ANETA MORKOVSKA, doctor, Czechia

"My name is Aneta Morkovska and I am 27 years old. I come from Czechia and I currently live in Prague. I have a rare disease, diagnose is called MRKH – MayerRokitansky-Kuster-Hauser syndrome, which is congenital aplasia of vagina and total agenesis of uterus. I had to undergo number of operations to have functional vagina. I basically do not experience serious health issues since that, but social and psychological impact is significant. About 2 or 3 years ago I decided to change my attitude and I joined the Czech Association for Rare Diseases as a first women with my diagnose and started to talk about it publicly. Czech Association for Rare Diseases helped me to spread awareness about the condition since then. I gave an interview to a countrywide magazine and to national radio station thank to them. I also try to educate medicine students and I make a presentation for them about my syndrome. I founded supporting group – including website and Facebook page – to connect women with the MRKH syndrome in my country. I usually meet with young girls/women or their relatives and tell them what to expect, how to deal with the condition itself and basically encourage them. In my personal life I finished university last year and I work as a doctor in Motol University Hospital in Prague and live with my boyfriend."

CATERINA DALLAI, psychology student, Italy

''My name is Caterina Dallai, I'm 22 and I've a bachelor's degree in psychology (earned at the University of Padova). At the moment I'm in a gap year between my degree and the beginning of a master, in social psychology. This year I'm a scoutmaster with kids from 8 to 12 years, and I've worked with kids (12-16 years), who have learning disabilities, to help them with homework. I've spent all the month of February in Bethlem volunteering at a school for hearing impaired children and teenagers. For two years, when I was in Padova, I volunteered as a hospital clown with a local organization (Dottor Clown). I've many hobby, such as playing guitar, taking photos and anything related to craft works (drawing, sewing and recycle item). In September I'll begin a master degree in social psychology, specifically the study course I'm interested about, study the communities and the promotion of well-being and social change."

DANIELLE DRACHMANN, executive director, Denmark

''My name is Danielle Drachmann. After giving birth to two children with idiopathic ketotic hypoglycemia (IKH) I took a Master in Anthropology of Health to support my passion for patient advocacy. Idiopathic ketotic hypoglycemia is a frequent cause of childhood hypoglycemia that is often underdiagnosed and undertreated. I founded the global organization Ketotic Hypoglycemia International (KHI) to foster the advancement of research into its etiology, prevalence, and treatment. From the very beginning parents and patients worked closely with the Scientific Advisory Board on various patient-initiated (research/publications) projects, based upon novel observations in the organization. Our work is an example of how a patient organization and medical experts can work together, connecting family representatives and experts from around the world. As the Executive Director of KHI, my work spans wide. From working on research protocols, medical publications, children's books, conveying research, coordinating conferences, fundraising, support patients in person and virtually, uniting the right experts around the different projects, approaching policy issues to establish and maintain the collaboration between pharma, biotech and IKH experts centralized around the patients in KHI.''

DOROTHEA DALIG, consultant in public affairs, Belgium

''My name is Dorothea Dalig. I studied psychology, Jewish studies, literature and pharmacy at the University of Freiburg, Germany. I graduated with a degree in psychology and pharmacy. I was actively involved in several form of students' representation and advocacy at faculty, national and international level focusing on access to education, students' participation, mobility and interdisciplinary approaches. 2017, I moved to Brussels to serve as Vice President of European Affairs for the European Pharmaceutical Students' Association (EPSA) and to work as a policy trainee for the Pharmaceutical Group of the European Union (PGEU). Since 2019, I work as a consultant for the public affairs agency Interel while practicing as a community pharmacist from time to time. My mother tongue is German, I am fluent in English and have a good command of French."

ELEONORE MATHE, pharmacy doctor, UK

''My name is Eleonore Mathe. French Doctor of Pharmacy, I chose to specialize in Regulatory Affairs after a first experience at Sanofi Pasteur, working on the Regulatory aspects of WHO/PAHO tenders for vaccines for Latin America, which also confirmed my interest in Public Health. After graduating from my International Drug Development and Registration MSc at

Paris Sud University, I moved to London to join GlaxoSmithKline where I had the opportunity to work at a global level in several therapeutic areas (Respiratory, HIV, Oncology, Rare Diseases). I then pursued my path at Spark Therapeutics to work in the development of gene therapies for Rare Diseases. These promising therapies being innovative, the regulatory landscape in this area has to adapt quickly. I feel strongly engaged in making the framework evolve and more supportive in order to facilitate patient access to innovative therapies.''

ELSA COULOMB, pharmacist, France

''My name is Elsa Coulomb and I am a French pharmacist. I decided to involve myself because I am afflicted by a rare disease. It is quite new, so I am still discovering what it to live with is. For now, I realize challenges that may be encountered, and I clearly realize that could be improved. So I want to become active in that way."

FANNI-LAURA MÄNTYLÄ, patient advocate, Finland

"Fanni-Laura Mäntylä is a Mental Health Activist, a Patient Advocate and Mental Health and Substance Abuse Work Professional. Mäntyla was Chair of the 5th Edition of European Health Parliament: Committee on Mental Health and Healthy Workforce.

Speaking openly about her personal experience of work related burnout and young adulthood with severe depression, Mäntylä in her 30s is now an active influencer about mental health in her native country Finland as well as in the EU.

Being a type-1 diabetic, Mäntylä urges for more discussion about health in a holistic manner and calls for all stakeholders together on the agenda. Mäntylä is a strong voice for young people affected by mental ill-health and a living proof that issues with health do not define a person."

HAYLEY HUBBERSTEY, health economist, UK

"My name is Hayley Hubberstey and I am from Wirral in North West UK, near Liverpool. I'm a science geek by background and now work in health economics specialising in understanding the impact of rare diseases to help enable access to new treatments. I have been involved with the rare disease community for about a year and it's been such a special experience. As part of my current job I've have the pleasure to attend and learn from many rare disease events and conferences. I felt instantly welcomed and at home as a professional new to the community. I have made new friendships and have met many inspiring and dedicated people. My education was in pharmaceutical sciences and I obtained my PhD in Biochemistry. I worked at Unilever a

global consumer goods company for 5 years. I joined as a laboratory scientist and then worked as a project manager. I gained experience in delivering successful projects whilst working alongside global cross functional teams. I managed these projects from project conception through to product launch. I now work for HCD economics as a real-world evidence manager. We use peer-reviewed methodologies to gather evidence to inform and challenge healthcare policies and to improve patient outcomes. We collaborate with a diverse group of healthcare professionals from academia, charities, patient advocates, clinical specialisms and industry sponsors."

MARIANA COUTINHO, clinical psychology researcher, Portugal

''My name is Mariana Coutinho. I have been diagnosed with a rare form of vascular sarcoma with a prevalence of 1 in 1 million and no standard treatment: Epithelioid Hemangioendothelioma (EHE). After being misdiagnosed, I was denied the possibility of accessing the most adequate treatment for me, which couldn't be provided in my home country. This was the moment when I decided to take my health into my own hands and started self-advocating. I learned about the EU Directive on Cross-Border Healthcare, one of the several legal instruments that allows patients to get access to treatment abroad, and became aware of my rights. Since then, I joined several patient associations where I work as a patient advocate and have had the opportunity to represent cancer patients on several national and European events. Recently I was invited to bring awareness and share my perspective at a roundtable event at the European Parliament where Cross Border Healthcare was discussed. For the last 2 years, I have also been doing volunteer work with children affected by cancer, and recently had the privilege of organising the meeting of a Portuguese MEP to this oncology centre. Additionally, I'm a Clinical Psychology Researcher and focus mainly on chronic illness."

MARIE BARTH, PhD biochemistry student, Germany

"My name is Marie Barth and I am a PhD Biochemistry student. I have been diagnosed with childhood cancer (Hodgkin Lymphoma). I am involved in various activities regarding childhood cancer: local survivor group, meetings on European level (child hood cancer international, SIOPe)."

ORIANA DE SOUSA, advisory board member, Portugal

"My name is Oriana and against all the odds, I am now a 9-year rare cancer (small cell ovarian cancer hypercalcemic type, aka SCCOHT) survivor due to immunotherapy. I started my journey

with cancer in 2011, at age 22, and found that the first response from my providers was one of ignorance and even negligence. However, my obstinate attitude and my parents' love enabled me to not give up; instead, I started self-advocating. I went through several hospitals in my country and abroad, always focused on finding a cure. After four recurrences, I became, in 2015, the first patient with my type treated with immunotherapy. I achieved total remission and my case let others follow the same path. Our results were encouraging to the research community and we worked together to understand when the good results were a random event or whether it would mean a new treatment option for our type of cancer. After seeing the results of the research, I gathered courage and accepted a New York Times interview. My goal was to not only be able to help more patients advocating for their treatments but also give visibility to my type of cancer and incite the medical community to look more closely for rare cancer patients and search for at immunotherapy and other personalised options. Once I am a Psychologist and, having in mind my personal life story, also feel the need to support emotionally and psychologically others who are dealing with cancer. For that reason, I recently founded a website for online psychological appointments to support cancer patients, survivors and caregivers. I am currently a student of the Patient Expert Course at the European Patients' Academy On Therapeutic Innovation (EUPATI) and I am Patient Advocate at Network Rare tumors of the Ovary (NRTO) and representative of a Patient Advisory Committee at EATRIS-Plus."

PASQUALE CACCIATORE, medical doctor, Italy

"My name is Pasquale Cacciatore. I am a Medical Doctor, currently finishing my residency program in Public Health at Università Cattolica del Sacro Cuore (Rome, Italy). During my residency I've worked on health policy, value-based healthcare, leadership in health and health economics. In 2019 I moved to Rotterdam (The Netherlands) to specialize in health economics, policy and law at Erasmus University. Since 2017 I have been the communication manager for EUPHAnxt, the network for students and young professionals of the European Public Health Association."

RACHEL CLAYTON, student in French and international relations, UK

"My name is Rachel Clayton. I am an undergraduate, studying French and international relations at the University of Leeds, UK. I am currently in my final year, working on my dissertation on rare disease policy in Europe alongside working with the team at the Leeds Human Rights Journal as their digital marketing officer. Through my family I have personally experienced the obstacles encountered by people living with a rare disease, as two of my family

members have rare diseases. I have supported through their journey in receiving diagnosis and treatment. My interest in rare diseases stems from the time I spent working at EURORDIS in the communications team and as part of the Rare Barometer initiative where I was introduced to their activities and the wider rare disease community. I look forward to learning more through the youth conference and contributing to the development of rare disease policy."

RITA FRANCISCO, PhD student, Portugal

"My name is Rita Francisco and I have a Masters' degree in Molecular Genetics by the University of Minho. I am currently doing my PhD focusing on better understanding immunological involvement in Congenital Disorders of Glycosylation (CDG) patients. My work is developed under the scope of the patient-led research network CDG & Allies – Professionals and Patient Associations International Network (CDG & Allies – PPAIN, http://www.researchcdg.com/). I am also a volunteer at the Portuguese Association for CDG (APCDG, http://www.apcdg.com), where I am the CDG patient advocate manager and CDG social manager. In 2018, I became an EUPATI Fellow and am an active member of EUPATI Portugal. I am also a member of the Patient Board at the European Reference Network for Rare Metabolic Diseases (MetabERN). I am passionate about science communication and believes patient-centricity can revolutionize research (and much more!)."

SEPPE KELCHTERMANS, policy advosir, Belgium

"My name is Seppe Kelchtermans. I am from Belgium and I am 26 years old. My background is in physical therapy and I specialized in neurological rehabilitation, more specifically, traumatic brain injury. Seeing how the patients I cared for and worked with faced significant societal hurdles when returning to society made me want to pursue a career in health policy, to try and impact the narrative around residual impairment and a move from being seen as a lifelong "patient" to being seen as a person again. In January of 2019 I joined the department of Economy, Science, and Innovation of the Flemish government as a policy advisor focusing on life sciences. I work on two, H2020 funded, CSAs. One on securing the uptake of personalized medicine in regions and the other one on the impact of the microbiome on human health. I also represent Belgium at the states representative group of the Innovative Medicines Initiative. During my work on personalized medicine the world of genomics opened up to me. As around 80% of rare diseases have a genetic component it struck me how much this field is still evolving. I am also a member of the European Health Parliament (EHP) committee on Europe as a Health Innovation Hub. I am excited to work together with you all and to learn more from people who have lived with a rare disease!"

STEFAN ZIVKOVIC, office assistant, Serbia

”My name is Stefan Zivkovic. I have an educational background in Political Science and International Relation, with a lot of non-formal education and training. I have volunteered/worked in various organisations in the field of youth, sustainable development and education (at both national and European level), involved in regional youth networks and have completed a traineeship in one of the political groups in the EP.

I work at the National Organisation for Rare Diseases of Serbia (NORBS), an umbrella organisation bringing together different rare disease patient organisations, with a main goal of providing a unified voice for all people affected by rare diseases and their family members, and representing them at different levels”.

THOMAS VILLARET, law student, France

“My name is Thomas Villaret, I am 23 years old, living in Avignon (France). I have a Diabetes Insipidus (a rare disease, totally different from Sugar Diabetes that you know) since I am 3 years old. At the very begging, that was very difficult to live with and I avoid death. I missed many class days so that was hard for me to make friends. There is a treatment for my disease but it was not balanced well. With time, things have been better. Finally, when I was 14 years old, I met specialists for this disease in Marseille and we found the good balance for the treatment. Now, I live almost normally (except the drugs that I have to take 3 times/day). I’m studying law in Montpellier like anybody else, actually I’m in master 1 (4 years). With time, this disease became a part of me and most of the time I forget totally the fact that I’m different from others. That’s why I don’t really like to talk about it, because it makes me remember something I don’t used to care about much than drinking or eating. Nevertheless, I am member of the AFDI (French association of Diabetes Insipidus) where I share advices with other members and I like to help new patients. I like video games, heavy metal and I have visited many countries like China, USA and European countries. I really enjoy talking with people and I learn at school to speak German, Italian and Chinese. My hobbies are medieval fencing (around 10 years now) and I play trumpet.”

JULIEN DELAYE, public affairs assistant, Belgium

“My name is Julien Delaye. I have joined EURORDIS in September 2019 and am now a Public Affair Assistant based in Brussels. My work within EURORDIS is now mainly focused on Rare2030, especially on the 2030 Young Citizen Conference. Prior to EURORDIS, I was a

volunteer for Osteogenesis Imperfecta Federation Europe (OIFE) and gained my first rare disease-related experience in Copenhagen, Denmark, where I did a marketing internship in a pharmaceutical company. I hold two bachelor degrees, in marketing (Liège – Campus Guillemins) and in European Public Health (Maastricht University), and a master degree in Public Policy and Administration (Dublin – UCD). Throughout my studies, I focused my academic work and papers on rare diseases, and my master thesis investigates the notion of significant benefit in Europe. I speak French, English, and bit of Dutch. My hobbies include traveling, photography, and poetry writing.''

KHADIDJA ABDALLAH, PhD researcher, Belgium

''My name is Khadidja Abdallah. I am a PhD researcher at KU Leuven (Belgium) and specialize in market access of orphan drugs with a focus on budget impact and payment approaches. I have a background in biomedical sciences, obtained my BSc and MSc degrees in resp. 2015 and 2018 from KU Leuven and have performed translational research on gene therapy at University College London. I am actively working towards delivering multidisciplinary and science-driven methodological guidance on optimization of payment approaches and reimbursement of orphan drugs to important stakeholders in Belgium. My recent review on methodological quality of budget impact analyses for orphan drugs has been presented at international conferences such as ISPOR and ECRD. Ultimately, I hope to contribute to the fair allocation of limited resources towards orphan drugs and promote sustainability in the treatment of rare diseases.''

ALESSANDRA BLONDA, Phd student, Belgium

''My name is Alessandra Blonda. After graduating in Drug Development at KU Leuven, I worked in the pharmaceutical industry for a couple of years. As my last position focused on market access & public affairs for biosimilars and generics, I became more aware of the issues regarding patient access to medicines, especially those for rare disease patients. For this reason, I went back to KU Leuven to start a PhD. Through my research, I aim to increase access to orphan drugs by optimizing existing reimbursement processes. By contributing to the Rare2030 project, I wish to become more involved in the rare diseases community; by learning from others, sharing ideas and supporting their common goal: improving the lives of rare disease patients, and making sure that no one is left behind.''

Annex 3: Fall Debate – Group presentations

Rare 2030
Foresight in Rare Disease Policy
GROUP 1

ALESSANDRA BLONDA
PASQUALE CACCIATORE
JULIEN DELAYE
DANIELLE DRACHMANN
SEPPE KELCHTERMANS
FANNI-LAURA MÄNTYLÄ

Young Citizen Conference
RECOMMENDATIONS

INTRODUCTION: IF WE HAD A MAGICWAND

> We would make sure that every rare disease patient has access to current, clear, and thorough expert information on their disease

> We would have equal access to healthcare for all patients irrelevant of location or socio-economic status

> All relevant actors would have the same access to reliable, ethical, patient-driven data

ESTABLISH A EUROPEAN RARE DISEASE AGENCY

> Overarching strategy
- Concentrating expertise, means, and resources
- Create a modular roadmap towards an integrated rare disease approach on EU level

> Focus on key actionable domains
- (Research) funding
- Social support
- Market access
- Data access and management
- Patient registries

RARE DISEASE AGENCY FOCUS AREAS– DIAGNOSIS

> Focus on newborn screening
- Minimum standard of hard to diagnose rare diseases included in newborn screening
- Comparative research on national newborn screening methodologies for best practice identification

> Foster joint-HTA assessment
- EU-wide initiative to assure access and availability of innovative, cost---effective, and safe treatment modalities

RARE DISEASE AGENCY FOCUS AREAS – POLITICAL AND STRATEGIC FRAMEWORK

> Clarify and reframe the role of ERNs
- Clarify how individuals can utilize this platform

> Reframe what it means to live with a rare disease
- Actively spread awareness to debunk stereotypes and stigmas

> Stimulate and increase public – private partnerships
- Fostering PPPs through incentives and other political means

CONCLUSION

> Focus efforts, expertise, funding through one EU-wide channel: The EU rare disease agency

> Focus on creating a community of experts, patients, scientists, policy makers that are engaged and willing to contribute

> Aim for co-creating and full value chain involvement from the ground up

> Move the knowledge not the patient

Rare 2030
Foresight in Rare Disease Policy

GROUP 2

Young Citizen Conference
RECOMMENDATIONS


ANA BEGIC


KHADIDJA ABDALLAH


RACHEL CLAYTON


AMINA BERBIĆ


DOROTHEA DALIG


ELEONORE MATHE

INTRODUCTION: IF WE HAD A MAGIC WAND WE WOULD …

> End discrimination towards people living with a rare disease

> Give equal opportunities and access to resources for people living with a rare disease

> Have equal access and availability to medicines and full reimbursement at European level

GROUP 2 | 2

RECOMMENDATION 1: DEVELOP A MORE COHESIVE AND MORE TRANSPARENT REIMBURSEMENT PROCESS

> **Why?**
- Individuals do not have a clear understanding of reimbursement procedures as they differ within countries and across Europe
- **Main actors: Insurers (private and public) , pharmaceutical industry, commission on reimbursement, minister of social affairs and health**

> **Possible actions**
- **Guidelines** created by the EU to encourage standardised processes across member states
- **Put a system in place that works when the state fails at european level –** database/ initiative that monitors patient medicines and treatments + reimbursement

> **Potential Challenges**
- data protection , transferring processes between localised and standardised , resistance against transparency from industry, patients and insurers (all countries have different systems)

GROUP 2 | 4

RECOMMENDATION 2: ADDRESS MENTAL HEALTH ISSUES AND SOCIAL CARE CHALLENGES

> **Why?**
- **Mental health issues often arise with living with rare diseases** and can be overlooked because of prioritising other treatment
- **The quality of life can decrease** if a person lives with mental health issues untreated, might exacerbate symptoms related to rare disease
- Mental health issues are becoming more prevalent in younger societies

> **Possible Actions**
- Inclusion of mental health aspect in treatment guidelines for rare diseases starting from diagnosis
- Training for mental health specialists on rare diseases

> **Potential Challenges**
- Not enough specialists, it is not necessarily a priority for doctors, cost

GROUP 2 | 5

RECOMMENDATION 3: TACKLE DISCRIMINATION VIA EDUCATION

> **Why?**
- Education on general health is not sufficiently emphasized.

> **Possible Actions**
- Training on disability and discrimination recommended to educators/ employers. Information more widely available.
- Awareness campaigns to reduce stigmatisation (Rare Disease Day)
- **Level of action:** European / national / regional

> **Potential Challenges**
- Rare diseases may get lost in the disability community, education may not be sufficient, awareness v action

GROUP 2 | 6

RECOMMENDATION 4: IMPROVE CROSS-COUNTRY COLLABORATION

> **Why?**
- **EU countries have different data collection standards**
- **Different care systems** with varying processes across borders

> **Possible actions**
- **Access to care abroad if needed –** a system at european level that pays health care professionals for the time they spend with **foreign** patients
- **Increasing the number of centres of expertise** to make them more accessible
- **Facilitating digital consultations** which include the patient, the GP, the specialist from a centre of expertise

> **Potential Challenges**
- **Challenges:** Member states' willingness to share resources, creating a harmonised process, what will incentivise the funding of a state to send a patient abroad, lack of specialists

GROUP 2 | 7

CONCLUSION

> **Common themes in recommendation:**
- **Collaboration / harmonisation**
- **Improved education and training**

> **Group experience/feeling**
- **Coming from different countries and different backgrounds but we come to similar conclusions**
- **There is a lot to do before we achieve scenario 3**
- **The initiative, working with young people, has opened our eyes to the possibility of change we can make**

GROUP 2 | 9

Rare 2030
Foresight in Rare Disease Policy

GROUP 3
ELSA COULOMB
STEFAN ZIVKOVIC
RITA FRANCISCO
THOMAS VILLARET
MARIANA COUTINHO
MARIE BARTH

Young Citizen Conference
RECOMMENDATIONS.

INTRODUCTION: IF WE HAD A MAGIC WAND

- We would have thriving partnerships between patients and other stakeholders (clinicians, researchers, policy-makers)
- We would have equal, borderless access to healthcare
- Patient data would be owned by the patient and stored safely (anonym)
- Patient-centred research would be the standard
- Every RD patient would get a quick and accurate diagnosis, and an effective treatment

GROUP 3 | 2

RECOMMENDATION 1: ENSURE THAT RD PATIENTS HAVE EQUAL ACCESS TO HEALTHCARE

- Why?
  - Inequalities between European countries in access to healthcare for RD patients
  - Differences between private and public sector
  - There is a need to optimize legal framework
- What for?
  - Accelerate communication/cooperation between european countries
  - Avoid delayed diagnosis/access to efficient treatments and care
  - Reduce the financial burden on RD patients and their carers
- How?
  - European guidelines for the management of rare diseases
  - Mixed funds for patients who need specialized treatments abroad
  - Improve the authorisation/payment process to access cross-border healthcare

GROUP 3 | 4

RECOMMENDATION 2: EDUCATION AND TRAINING OF ALL STAKEHOLDERS TO IMPROVE PARTNERSHIPS

- Why?
  - Foster knowledge and partnerships involving patients
  - Reduce cultural differences and barriers
- What for?
  - Better communication of and focus on patients and stakeholders needs
  - More efficient research
- How (if applicable)?
  - Training for patients, clinicians, researchers and policy-makers
  - Expert patients
  - Common EU medical school program to train doctors (patient rights, cross-border health care)

GROUP 3 | 5

RECOMMENDATION 3: FACILITATE DATA OWNERSHIP AND PROTECTION

- Why?
  - More digital data is accumulated (big data, EHR)
  - Sensible patient data
  - Access to health care
- What for?
  - Allow more data to be stored safely
  - Accelerate research and make it more efficient
- How (if applicable)?
  - Robust standards for legally and ethically data sharing
  - Patients as legal data owners (decide sharing parties)
  - App for authorisation of personal data use in specific cases

#my_data_my_choice!

GROUP 3 | 6

YOUR PERSONAL CONCLUSION

This experience has been:

"Demanding…Unique… the ultimate ACT now, CHANGE tomorrow exercise" - Rita

"This is a prime example of what it means to 'be ahead of the game' and how we are more prepared for what is coming if we stand together and have an active role in the decisions that concern us. I am truly grateful for everything I learned with this enriching experience" - Mariana

"Innovative Idea-Factory to improve the future of rare disease patients" - Marie

GROUP 3 | 8

Rare 2030
Foresight in Rare Disease Policy

GROUP 4

CATERINA DALLAI, Psychology Student, Italy
ORIANA DE SOUSA, Patient & Psychologist, Portugal
HAYLEY HUBBERSTEY, Real World Evidence Manager & Patient advocacy group chair, UK
ANETA MORKOVSKA, Patient & Doctor, Czechia

Young Citizen Conference
RECOMMENDATIONS

INTRODUCTION:

> DIAGNOSTICS:

> Shift from relative ignorance of the issue towards general awareness of the diagnostic issue in RDs (awareness amongst doctors)

> INTERGRATED, SOCIAL, HOLISTIC CARE:

> Recognising the role of health care providers and other social services (particularly those involved in e.g. European Reference Networks) in achieving these goals

RECOMMENDATION 1: DIAGNOSTICS

> Why? Issue & groups involved

- ISSUE: Cost of developing test, Doctors skills to dedicate, not enough incentives, rolling out tests and communication
- GROUPS INVOLVED: Healthcare system from local hospitals to international networks

> What/how?

- RECOMMENDATION: Training and education of networks of doctors on the availability of test and devices provided in conjunction with patient associations
  - Patient associations to promote partnerships with academic institutions programme. Working with academic institutions for training for health care professionals making sure rare disease is on the teaching agenda early on
  - Organisation of annual virtual meetings for key groups and doctors, increased coordination between ERNs and hospitals to develop protocols and flowchart of symptoms to consider rare diseases
- LEVEL OF ACTION: National and European
- CHALLENGES: Low cost but high efforts in coordination and continuing to get the message out

RECOMMENDATION 2: INTERGRATED, SOCIAL, HOLISTIC CARE

> Why?

- ISSUE: Doctors don't often assess non clinical needs/daily needs as they are already very busy with their schedule and unaware of the intricacies of rare disease
- GROUPS INVOLVED: Case managers, Social workers, Doctors network, Psychologists, Patient organisations, Patient advocates & experts, other HCPs, Academics & Health economists.

> What/how?

- RECOMMENDATION: Bring social care into the healthcare system by creating coordination type roles to bridge the gap within countries and across the EU. This is followed up with research on holistic care to generate evidence of importance for ongoing support.
  - By sharing studies which already exist and have worked well on a national level and lobby to move to EU level to gain funding.
  - Expand the scope of ERNs to include both integrated and social holistic care and include a larger group of stakeholders all together
  - Continue to talk and include patients and caregivers to fully understand the needs
  - Raise awareness of the resources already available and work with rare disease organisations
- LEVEL OF ACTION: National and European
- CHALLENGES: High cost, might be difficult to convince governments but the data generated in the long term will support its worth

CONCLUSION

> Common themes in recommendation

- Multi-disciplinary teams
- Keeping the patient voice, patient experts and patient associations centre front
- Better use of ERNs or creating central clinics/units for rare disease in Europe (one stop shop) working with or in hospitals directly
- Providing data or education to continue learnings more about rare disease and the benefits of this work
- Cost is always a barrier and find solutions around this, the cheaper less perfect options, or joint solutions where countries can work together to share the costs.
- A need for providing evidence on the benefits vs cost of a new approach vs long term cost of not implementing anything, a need to work with academics and health economists on generating data.
- Doctors and HCPs time is always a barrier so lets make things as straight forward as possible and keep sending the message out

> As a group we really enjoyed working together and we all had different experiences. We had patients, students, doctors and researchers in our group all from different countries in Europe. This enabled us to form well considered and well formed ideas.

> We identified a key theme of the doctor-patient model and a paradigm shift in communication because a patients quality of life is more than just the clinical aspects of the condition

Annex 4: Young Citizens Open Letter

Dear Members of the European Parliament, dear European Commissioners, dear present policy makers and leaders,

You have done tremendous work in the past decade to improve the lives of people living with a rare disease in Europe. However the world we live in today is different than the one we knew back then. With the emergence of new contexts, challenges, but also new opportunities, we need further concrete actions to guarantee that Europe is indeed a place where no one, including people living with a rare disease, is left behind. It has taken a generation to get where we are. Now it's time to listen to what we - the Rare 2030 Young Citizens and the next generation of health and social care providers - have to say about the future.

In ten years' time, I will still be living with a rare disease. I may also be a parent of a child living with a rare disease. In ten years' time, we will have lost too many of our peers, too early, to rare diseases. By 2030, I want to live in an inclusive society that recognises all my needs and acknowledges that my condition goes beyond physiological symptoms. I want to have access to the right diagnosis quickly and to be given the appropriate and effective treatment options wherever I live and whatever my socio-economic status may be.

In ten years' time, I will be a doctor, I will be a nurse, a psychologist. By 2030, I want to work and collaborate with colleagues in my country and throughout Europe who have solutions when faced with rare diseases, to whom rare diseases are not a mystery. I want us, health and social care providers, to become the best version of our professions with the tools and mindsets needed to make sure that our patients can live the healthiest and happiest life they can.

In ten years' time I will be a policy maker. By 2030, I want rare diseases to be seen as a public health priority. I want Europe to be an example of fairness, equity and inclusiveness in all areas having an impact on the lives of people living with a rare disease. I want the benefits of cross-border collaboration to be recognised and obvious - no longer a proof of concept.

We, the Rare 2030 young citizens, come from different backgrounds, different countries and have different experiences with rare diseases. We are the next generation of patients, patient advocates, doctors, nurses, psychologists, policy advisors and policy makers. We want to be the generation of change. As the leaders of today you can make a meaningful difference, to drive change for our generation, and the generations of rare disease patients, carers, doctors and policy makers to come.

Dear Members of the European Parliament, dear European Commissioners, dear

present policy makers and thought leaders, **please don't let the health and well being of 30 million people living with a rare disease in Europe be left to luck or chance**. Consider our words in your policy changes today.