



Asia Pacific Alliance
of Rare Disease Organisations

REPORT FROM ECRD and RDI Virtual Conferences

15-19 MAY 2020

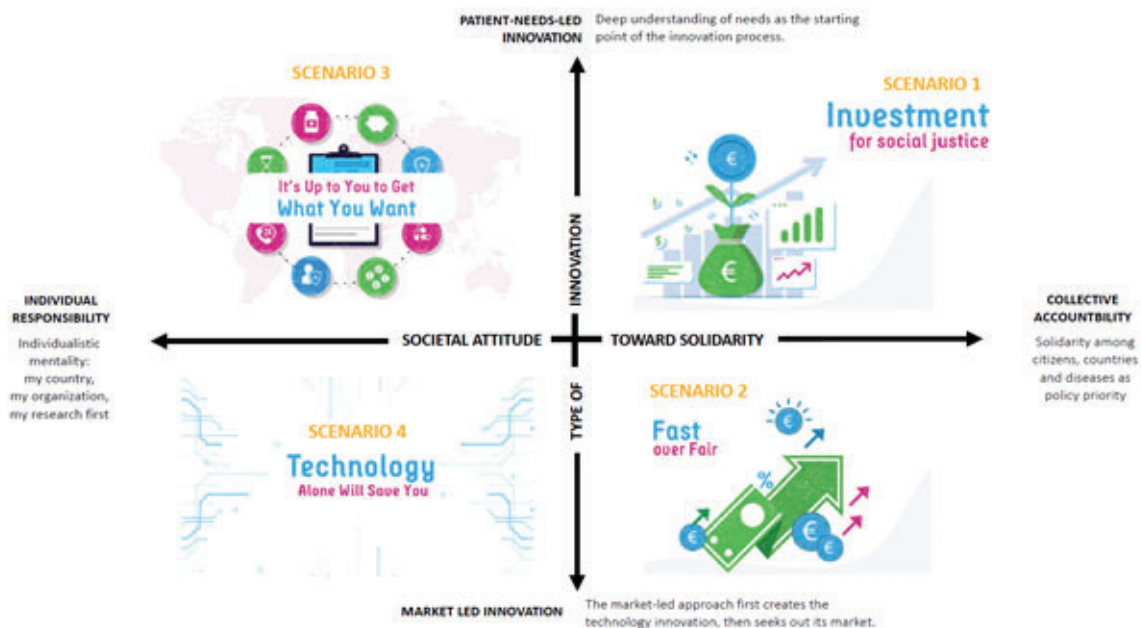


BY: LISA FOSTER, 12 JUNE 2020

The ECRD is recognised globally as the largest, patient-led rare disease event in which collaborative dialogue, learning and conversation takes place, forming the groundwork to shape future rare disease policies. One of the opening statements by Stella Kyriakides, European Commissioner for Health & Food Safety summed up the conference ethos,

"In the coming years we are carrying out a real stocktaking exercise, we will be guided by the Rare2030 Foresight Study. We need now to take lessons from COVID-19. We know that patients will be the driving force of our rare disease policy."

Rare 2030 Foresight Study is a 2-year Eurordis led study to propose policy options leading to 2030 that will provide a better potential future for people living with rare disorders. This has been formulated with extensive input from over 200 experts in varied fields and shortened into four quadrants. The possible scenarios/quadrants presented are expressed in this diagram: -



Following Covid-19 there was a reflection on where we are currently and which scenario we hope to move towards and how we might get there. The learning that a single health issue such as a virus has the power to block entire global economies was discussed as it highlights the intricate link between healthy societies and healthy economies. The four scenarios were presented and discussed with the collective patient organisation focus on reframing of public health policies towards 2030. From this the outcome was ideally the world can move toward a new paradigm with a shift towards inclusion and investment for social justice scenario or model.

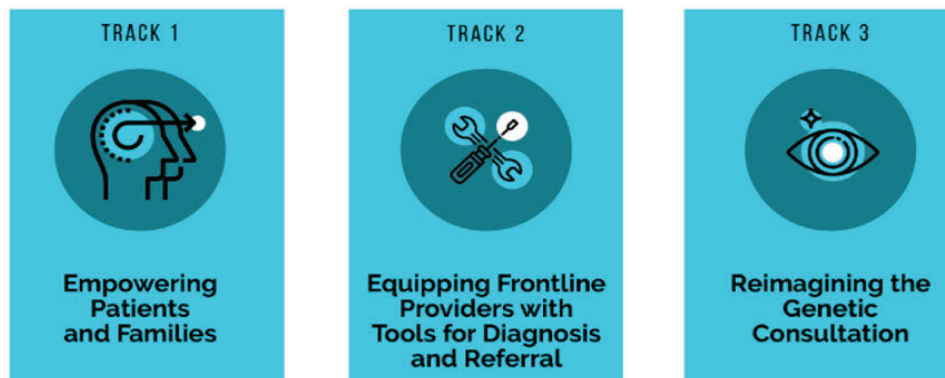
As part of this the need for collective voices, collaboration to strengthen all countries to head in the same direction and improved diagnosis was discussed. Practical opportunities to connect globally to help with a better future for all people with rare diseases was highlighted with some of the following focus areas: -



- Global commission to end the diagnosis odyssey for children with a rare disease – one year report and opportunities for APARDO countries to become more involved to gain from inclusion in this collaboration <https://www.globalrarediseasecommission.com/>

Global Commission Year One Report

The Global Commission Identified three key pathways following the group's research:



Projects include **Dx-29** and Solve-RD which is a tech pilot currently being tested in Spain which utilised AI to recognise symptom patterns to inform and guide health practitioners and so far, has **79%** success rate (from GP's). It also links to telehealth and facial recognition to allow a great referral process to geneticists.

Some other highlights included: -

- Rudiger Krech, Director of UHC provided support and oral statement on needs of persons with disabling Rare Diseases
- Discussion on the benefit and use of the current Memorandum of understanding (Dec, 2019) with World Health Organisation for Collective Global Networks to strengthen competency across the world and a Needs Assessment and an operational description of rare diseases.

<https://www.rarediseasesinternational.org/rdi-signs-memorandum-of-understanding-with-the-world-health-organization/>

The activities that will be carried out as part of this MoU fall within a broader longer-term strategy that lays out the following ambitious goals:

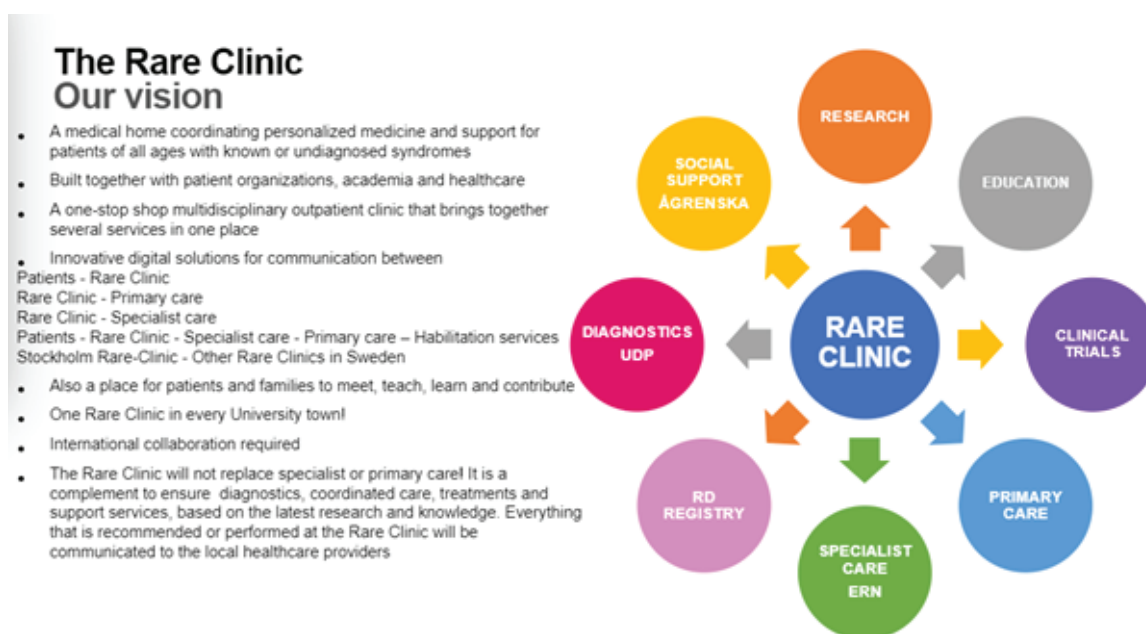
1. Increase awareness of rare diseases and their visibility in healthcare systems;
2. Strengthening healthcare systems, including improving access to accurate diagnosis, prevention of co-morbidity, and promotion of multidisciplinary holistic care;
3. Improve access to affordable and quality treatments and care; and
4. Encourage Member states to adopt strategies to address rare diseases challenges at national level within an international policy framework.

- Digital Innovation Patient networks are vital for data analysis and there are ongoing efforts to make sure that patients are driving positive changes to boost rare disease data analysis- there is the benefit of having set global standards within 'Go Fair' data principles which were set up in 2016.
<https://www.go-fair.org/fair-principles/>
- Citizen driven policies - Patient Journey shaping clinical journeys (Olivia Spivak). This session focused on how the European Regional Networks (ERN) pull together expertise from the patient groups to inform and support the healthcare professionals. They used a patient journey visual template table to help guide the process and facilitate discussions with clinicians in a structured way.

Many discussion about Human Rights, Disability and Advocacy- there was a focus on Scandinavian countries who have national advisory units on rare disorders (Norway) and have a joint view with all of those with complex medical needs so not to be siloed and separated but to work together on common areas with a collective voice for the wider disability movement. As the social model of disability includes impairments but often creates barriers to access for rare 'invisible' disorders due to system failures people can be left behind. The fact that labels can cause separation and division was made and a solution to act more collaboratively with an empowerment perspective including wider society was proposed. As stated by **Rebecca Tvedt Skarberg**

Disease/Disability of a person should not make any difference, Because Inclusion means supporting the functionality of a person to enable reaching the best of his/her full potential

- A Swedish model was presented on Rare Clinics <https://rarecare.world/> as displayed in this diagram: -



Rare Barometer Survey breakout room- 5,000+ rare disease patients and their family members from all countries representing **993 diseases** responded to the survey carried out via the Rare Barometer Programme. The analysis is still to be clarified however the main aspects show

- 9 in 10 people had an interruption in care with 6 in 10 of those declaring an interruption of care related to the COVID-19 pandemic was detrimental to their health or the health of the person they care for
- More than a half of those who need surgery or transplant have seen these interventions cancelled or postponed
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- 3 in 10 perceive that these interruptions of care could definitely (1 in 10) or probably (2 in 10) be life-threatening
- Patients who usually receive care in hospitals are experiencing specific difficulties, with almost 3 in 10 reporting that the hospital or unit that normally provides care for their rare disease is closed
- 1 in 2 have participated in online consultations or another form of telemedicine since the start of the pandemic. This is new for 2 in 10 patients. Almost 9 in 10 of those who have experienced this type of consultation are happy with the experience and that it has been very or fairly helpful

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