

3rd Conference of International Rare Diseases Research Consortium

Paris, France
8 – 9 of February, 2017

Anders Olauson

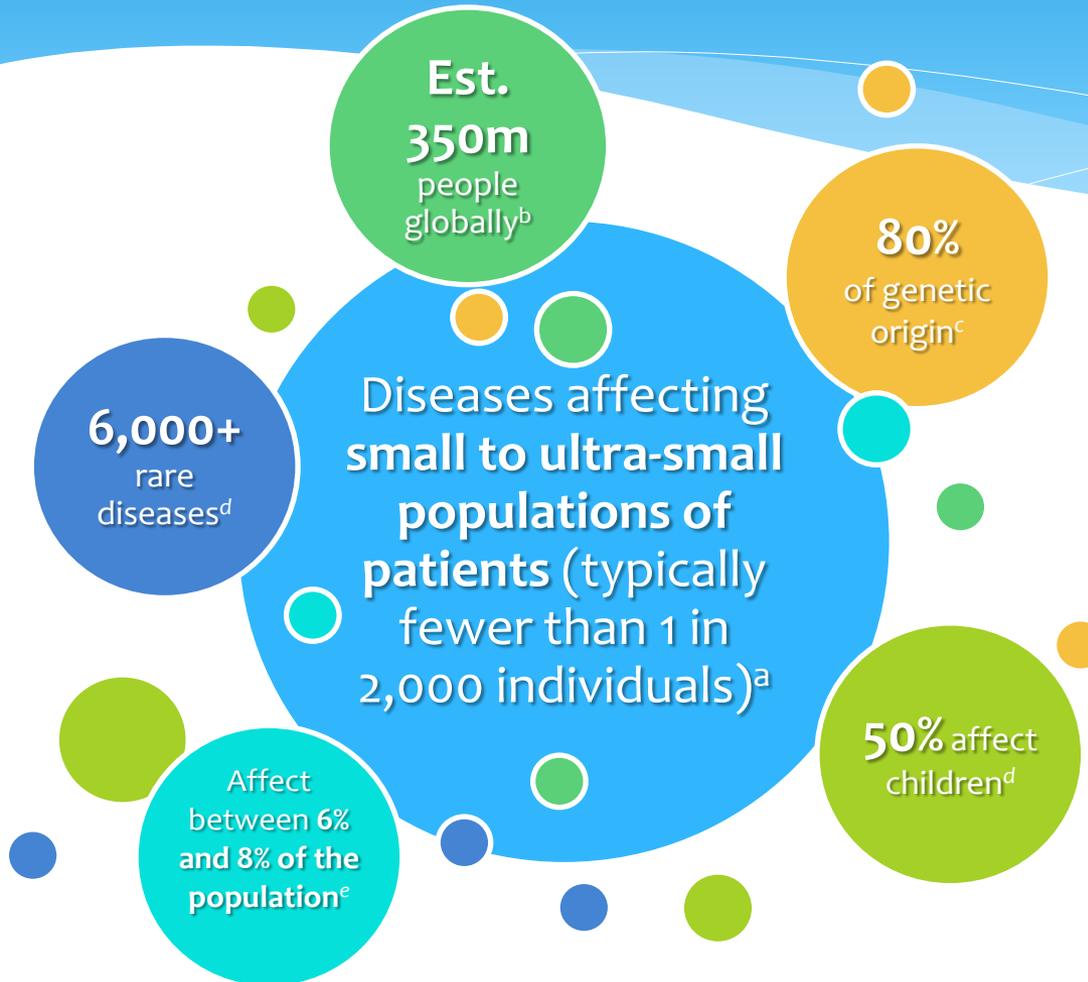
A Committee of **CoNGO**



NGO COMMITTEE FOR
RARE DISEASES



What are rare diseases?



^a European threshold as defined in EU Orphan Drug Regulation 141/2000. Thresholds in other regions/countries may vary.

^b Estimate based on statistics available across different populations.

^c http://www.who.int/medicines/areas/priority_medicines/BP6_10Rare.pdf

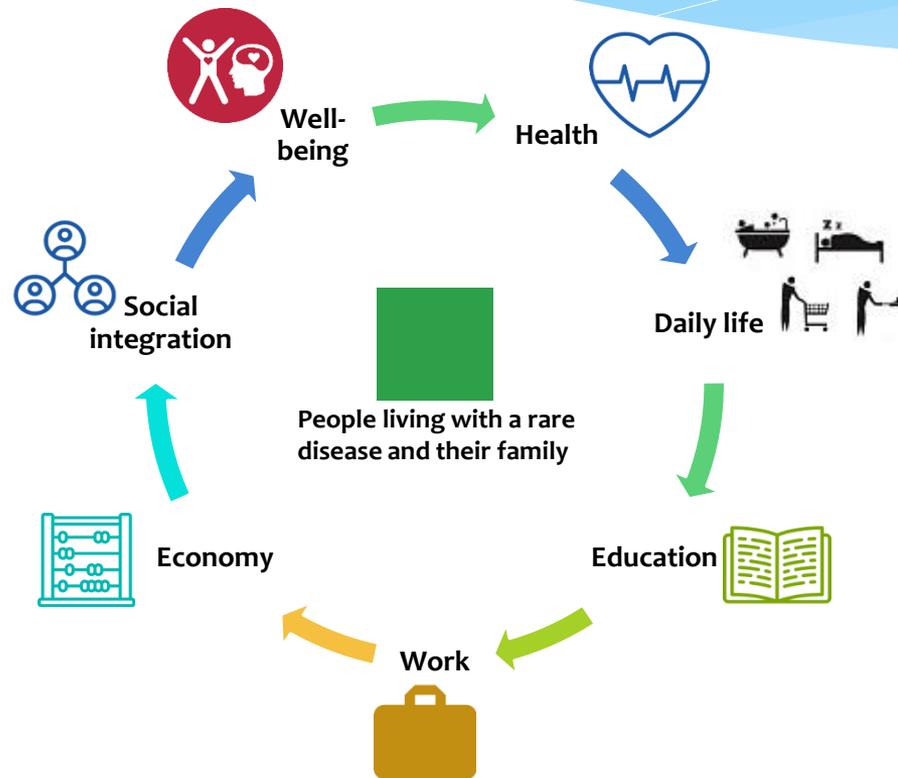
^d http://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN

^e Council Recommendation of 8 June 2009 on an action in the field of rare diseases. Paragraph 5. Downloaded from: [http://eurlex.europa.eu/LexUriServ/LexUriServ.do?uri=CELEX:32009H0703\(02\):EN:NOT](http://eurlex.europa.eu/LexUriServ/LexUriServ.do?uri=CELEX:32009H0703(02):EN:NOT)

Hurdles in Care Provision

- * People living with a rare disease need to be followed simultaneously and continuously by a set health, social and support services;
- * These are often managed by different authorities/providers and there is a lack of communication and coordination between them;
- * Care systems are fragmented and extremely difficult to navigate for patients and families;
- * Professionals lack knowledge on RDs and tend to be reluctant to treat patients due to the complexity of their disease;
- * In most cases, the management and coordination of care has to be done by patients and families, which places a heavy burden on family life.

A rare disease can affect many spheres at once



FRIENDS

FAMILY

SIBLING

SCHOOL



HOSPITAL

PARENTS

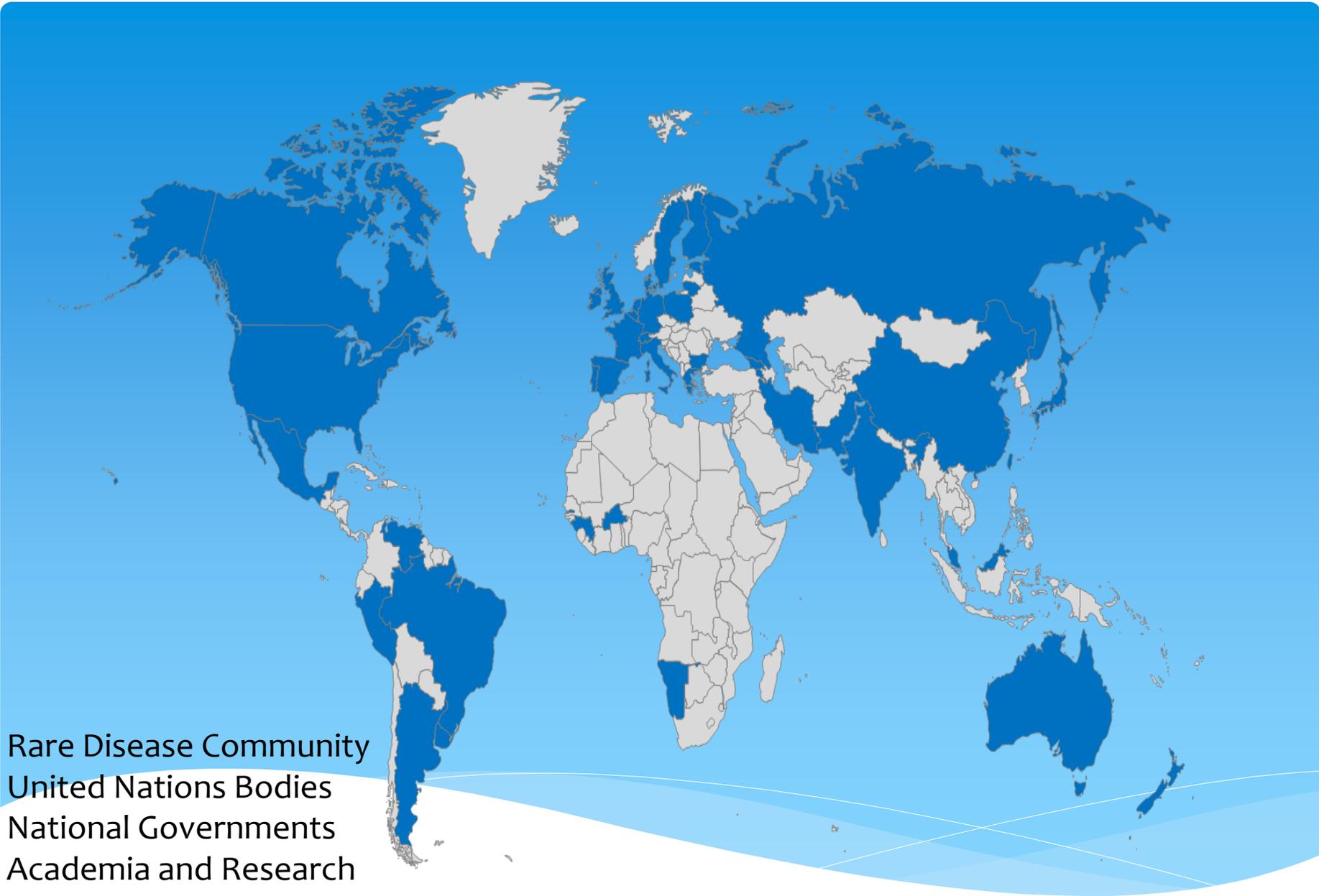
UN NGO Committee for Rare Diseases





Partnes and oldies
across all borders.





Rare Disease Community
United Nations Bodies
National Governments
Academia and Research
Pharmaceutical Industry
Other Civil Society Organisations

Founding Act

« Rare Diseases and the UN Sustainable Development Goals »

New York City, Friday 11 November 2016

We, the undersigned participants to the inaugural « Global Gathering for Rare Diseases », proclaim and endorse the following statements:

- Each of the 6,000 repertoried rare diseases affects a very small population locally. **All together, however, they represent an international public health issue.**
- The 350 million persons living with a rare disease worldwide are facing **common challenges across diseases and across borders** that affect all parts of their lives at once.
- No one country, no one continent alone can solve the problems posed by rare diseases.**
- Experience shows that actions are possible and effective. **Common national policies and international collaboration can address these challenges.**
- Scientific, medical, technological, social opportunities will be high in the next 20 years. **Patients, NGOs and other stakeholders are committed and aligned for partnership.**
- Rare diseases fit within the objectives from several UN SDGs  and can significantly contribute to their achievement.

The NGO Committee for Rare Diseases will catalyse all efforts towards the delivery of the SDGs in support of rare diseases, and towards the recognition and integration of rare diseases in all relevant future global policy and initiatives of the United Nations and its agencies.



How and in which fora

In order to do this, there are various fora and initiatives available to rare disease NGOs that are in consultative relationship with the United Nations, including those groups that are members of the NGO Committee for Rare Diseases:

[The High-Level Political Forum \(HLPF\)](#): this is United Nations' central platform for the follow-up and review of the 2030 Agenda for Sustainable Development and the SDGs. The HLPF includes voluntary country reviews, thematic reviews on progress of SDGs as well as a range of side events that can be organised by major stakeholders like civil society.

[ECOSOC Partnerships Forum](#): Every year ECOSOC and other UN entities collaborate to bring together leaders from government, the private and non-profit sectors and civil society to discuss multi-stakeholder partnerships.

[The World Health Assembly](#): This is the decision-making body of the WHO and determines the policies of the organisation, appoints the Director-General and approves the proposed programme budget. There are also a number of side-events organised on the same days.

Official International days: These can be a good opportunity for strong communication and advocacy campaigns such as [Official WHO health days](#).

[The Commission for Social Development \(CSocD\)](#): This is the key UN body advising ECOSOC on social policies of a general character and, in particular, on all matters in the social field not covered by the specialised inter-governmental agencies.

[The UN Convention on the Rights of People with Disabilities \(CRPD\)](#): This is intended as a human rights instrument with an explicit social development dimension. As it adopts a broad categorization of persons with disabilities it is possible for the rare diseases community to employ this instrument in its advocacy where relevant and hold accountable those countries that are signatories.

[UN General Assembly Resolutions](#): These are voted on by all member states of the UN in the General Assembly. They usually require a simple majority to pass. These are generally non-binding towards member states but they may act as soft law. Rare Diseases advocates can try to collaborate with governments to push for a resolution on Rare Diseases (particularly on Rare Disease Day as it was done for Diabetes).

The need for global action against rare diseases

No one country, no one continent, can solve alone the problems posed by rare diseases.

- * Numbers of people living with rare diseases as well as medical and other experts, even at a continental level, are too low and geographically scattered to reach the point of good health and well-being for every person living with a rare disease
- * A critical mass of people living with rare diseases as well as other experts and public health authorities must be brought together internationally
- * People living with rare diseases still remain a marginalised, vulnerable and largely invisible population within healthcare and social systems
- * There is a need to develop and implement definitions of rare diseases in many countries. This is a pre-condition for both national awareness, knowledge and international comparisons.
- * With little or no awareness and understanding of the needs of people living with rare diseases, they are often left unmet



The globalisation of rare diseases is gearing up...

* Research:

- * IRDiRC: an International Rare Disease Research Consortium
- * International platforms for rare disease registries

* **Patients** are increasingly organised across borders.

* Policy:

- * Rare disease policy, strategy and plans implemented nationally, having a 'snowball' effect in other countries
- * Incentivizing policies for industry investment e.g. US Orphan Drug Act and EU Orphan Medicinal Products Regulation
- * Increased collaboration between agencies (EMA, FDA) + EU-WHO Data Sharing Pact



More needs to be done

There is an urgent need for a **global platform**:

- * To share the scarce knowledge that we already have: it is applicable, valid and useful for all socio-economic and cultural contexts.
- * To explore what more could be done or developed to advance knowledge of rare diseases at a global level.
- * To connect rare disease stakeholders across borders and diseases.
- * To create synergies with other stakeholders to mutually exchange knowledge and expertise.
- * For recognition and attention at the UN level, where rare diseases remain an area little explored, with great social and economic impact.

Common

Goals:

Why is the United Nations the right platform?



NGO COMMITTEE FOR
RARE DISEASES



Why the United Nations?



World Health
Organization



 **UN
WOMEN**

The right place...
For decades, the UN has been a driver of a strong public health agenda, social development, inclusion, and progression, developments in science, technology and innovation, and human rights for all.

unicef 



United Nations
Department of Economic and Social Affairs



ECOSOC
United Nations

Why the United Nations?

... at the right time

In November the UN adopted its **2030 Agenda for Sustainable Development**, including the adoption of the 17 **Sustainable Development Goals (SDGs)**.

There is a clear alignment of a number of goals with rare diseases, e.g.:



1 NO
POVERTY



GOAL 1: END POVERTY IN ALL ITS FORMS EVERYWHERE

1.3

- * Implement nationally appropriate **social protection systems and measures** for all, including floors, and by 2030 achieve substantial coverage of the poor and the **vulnerable**.

1.4

- * By 2030, ensure that all men and women, in particular the poor and the **vulnerable, have equal rights to economic resources, as well as access to basic services**, ownership and control over land and other forms of property, inheritance, natural resources, appropriate new technology and financial services, including microfinance

- Exclusion from health and care systems
- Lacking social protection
- Inaccessible educational environment
- Lower employment rates
- Absence from work due to inaccessible labour markets for people living with rare diseases and family members
- Expensive treatments, special resources and adaptations
- Inability to live in dignity and autonomy with no income and costly adaptations to be autonomous

3 GOOD HEALTH AND WELL-BEING



GOAL 3: ENSURE HEALTHY LIVES AND PROMOTE WELL-BEING FOR ALL AT ALL AGES

SDG Targets

- * **3.2**
 - * By 2030, **end preventable deaths of newborns and children under 5 years of age**, with all countries aiming to reduce neonatal mortality to at least as low as 12 per 1,000 live births and under-5 mortality to at least as low as 25 per 1,000 live births
 - * **3.4**
 - * By 2030, **reduce by one third premature mortality from non-communicable diseases** through prevention and treatment and promote mental health and well-being
 - * **3.8**
 - * **Achieve universal health coverage, including financial risk protection, access to quality essential health-care services and access to safe, effective, quality and affordable essential medicines and vaccines for all**
 - * **3.b**
 - * **Support the research and development of vaccines and medicines for the communicable and non-communicable diseases**
- Poor understanding of rare diseases and their needs as patients and experts are few and geographically scattered.
 - Slow or misdiagnosis
 - Highly complex, often degenerative and disabling
 - Life expectancy is often severely affected
 - Only 5% of rare diseases have an adequate treatment
 - Unaffordable and inaccessible treatments
 - Profound psychological burden
 - Preventable deaths through newborn screening
 - Lack of incentives to boost research and development of rare disease treatments.



GOAL 4: ENSURE INCLUSIVE AND EQUITABLE QUALITY EDUCATION AND PROMOTE LIFELONG LEARNING OPPORTUNITIES FOR ALL

SDG Targets:

- * 4.2
 - * By 2030, ensure that all girls and boys have access to **quality early childhood development, care** and pre-primary education so that they are ready for primary education
 - * 4.5
 - * By 2030, **eliminate gender disparities in education** and ensure equal access to all levels of education and vocational training for the vulnerable, **including persons with disabilities**, indigenous peoples and children in vulnerable situations
 - * 4.a
 - * Build and upgrade **education facilities that are child, disability and gender sensitive** and provide safe, non-violent, inclusive and effective learning environments for all
- *50% of rare diseases affect children*
 - *Difficulties to attend school and university and to develop in a quality manner due to rare diseases:*
 - *Negative impact on health*
 - *Frequent medical appointments*
 - *Inaccessibility of facilities*
 - *Inaccessible teaching methods*
 - *Ineffective translation of medical information to teachers and educators who do not understand what the rare disease is, nor the impact it has and the needs it creates, limiting quality education*



GOAL 5: ACHIEVE GENDER EQUALITY AND EMPOWER ALL WOMEN AND GIRLS

SDG Targets:

- * **5.1**
 - * End all forms of discrimination against all women and girls everywhere
 - * **5.c**
 - * Adopt and strengthen sound policies and enforceable legislation for the promotion of gender equality and the empowerment of all women and girls at all levels
- **Primary carer role** for people living with rare diseases is mostly **assumed by the mother** ([ENSERio](#))
 - Mothers of children with a rare disability reported **high parental stress** and **high physical and emotional strain** ([Delve et al, 2006](#))
 - **Pain is considered less seriously in women** than in men ([EURORDIS Care](#)) e.g. Ehlers Danlos, CF
 - **Women face more rejection in looking for a diagnosis** ([EURORDIS Care](#)), meaning diagnosis takes longer and treatment is delayed e.g. Crohns disease, Cystic Fibrosis
 - **Gender equality and women in science**



GOAL 10: REDUCE INEQUALITY WITHIN AND AMONG COUNTRIES

SDG Targets:

* **10.2**

* By 2030, empower and promote the social, economic and political inclusion of all, irrespective of age, sex, disability, race, ethnicity, origin, religion or economic or other status

* **10.3**

* Ensure equal opportunity and reduce inequalities of outcome, including by eliminating discriminatory laws, policies and practices and promoting appropriate legislation, policies and action in this regard

- *Health inequalities and disparities between regions in acknowledging rare diseases*
- *Invisibility of rare diseases within health care systems of LMIC*
- *Inequality in accessing health care services, and inequality of treatment and integration compared to more common diseases*
- *Worse quality of life and experience higher losses in terms of medical care and social and economic activities compared to more common diseases (Van Nispen 2003)*
- *Inequality in provisions of and price of treatments.*
- *Discrimination in the labour market*



GOAL 17: REVITALIZE THE GLOBAL PARTNERSHIP FOR SUSTAINABLE DEVELOPMENT

Capacity building

- * 17.9
- * Enhance international support for implementing effective and targeted capacity-building in developing countries to support national plans to implement all the sustainable development goals, including through North-South, South-South and triangular cooperation

Multi-stakeholder partnerships

- * 17.16
- * Enhance the global partnership for sustainable development, complemented by multi-stakeholder partnerships that mobilize and share knowledge, expertise, technology and financial resources, to support the achievement of the sustainable development goals in all countries, in particular developing countries
- * 17.17
- * Encourage and promote effective public, public-private and civil society partnerships, building on the experience and resourcing strategies of partnerships

- *Rarity requires global collaboration*
- *Multistakeholder partnerships are necessary: public, private and civil society groups*
- *Patients and patient groups to be seen as partners*
- *The role of science, technology and innovation as an enabler of the post-2015 development agenda: rare diseases research drives innovation often beneficial to larger population groups.*

The NGO Committee for Rare Diseases



NGO COMMITTEE FOR
RARE DISEASES



Leave no one behind

Introducing the Committee



NGO COMMITTEE FOR
RARE DISEASES

Mission

Promoting rare diseases on the global level

Vision

An inclusive, multi-stakeholder ecosystem to share knowledge and expertise about rare diseases and to increase global visibility of rare diseases

Introducing the Committee

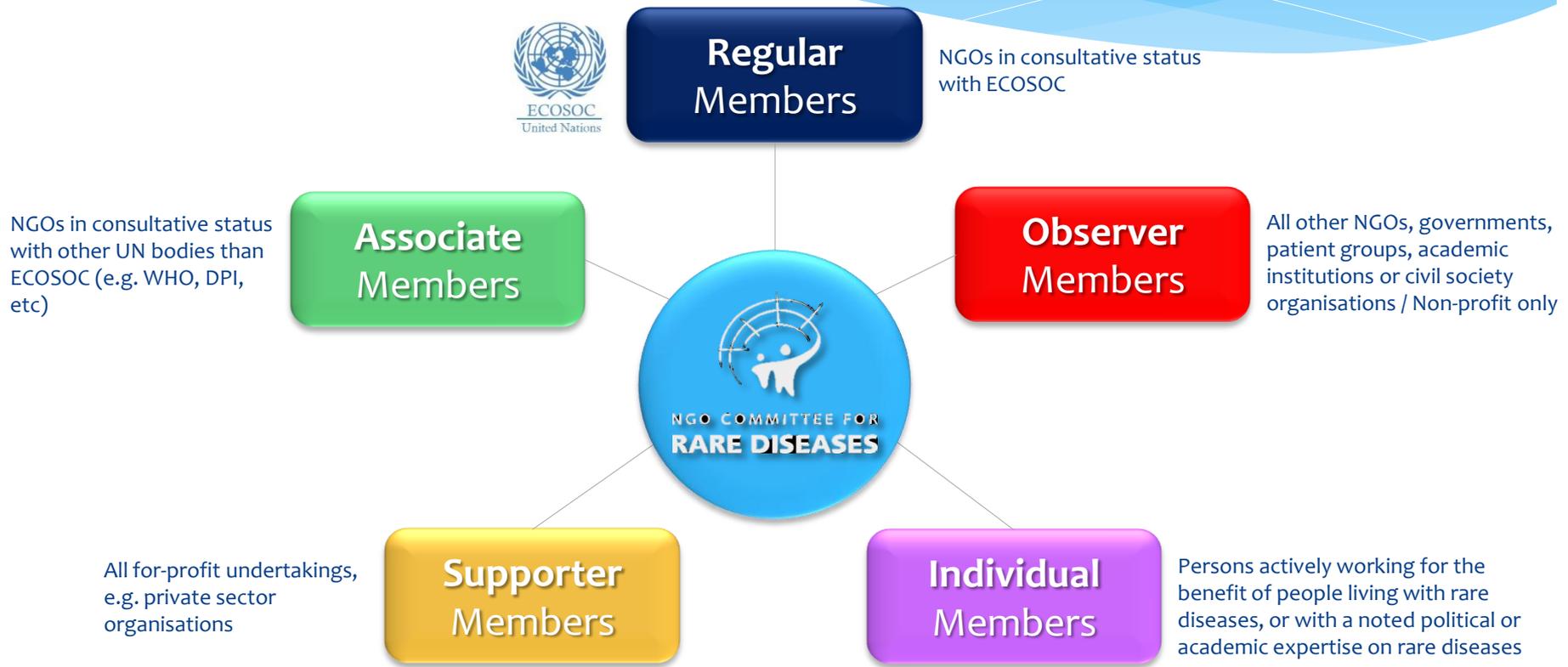
Objectives:

- * To **increase visibility** of rare diseases at the global level
- * To **extend and share knowledge** about rare diseases and their unmet needs
- * To **connect** NGOs interested in rare diseases and their partners within a global platform
- * To **promote international, multi-stakeholder collaboration and actions** for rare diseases
- * To **align** rare diseases as a global priority in public health, research and medical and social care policies

Potential deliverables

- * Two working meetings a year – one in Geneva (Spring), one in New York (Autumn)
- * A Rare Diseases Atlas: capturing the landscape of rare disease information, policy and initiatives around the world.
- * Short annual reports on specific issues of relevance to rare diseases
- * Publications, media articles
- * **... and more broadly, direct input into relevant policy initiatives or resolutions to be discussed within the UN system, at various levels (ECOSOC, WHO, UNESCO, IMF etc.)**

Introducing the Committee: Composition and Members



Our current timeline

October 2015



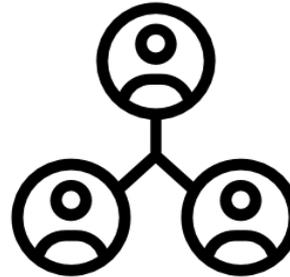
**Accepted
as a CoNGO
Committee**

March 2016



**By-laws
approved by
CoNGO
president**

April 2016



**Establishing
contacts
within UN
and
member
states**

November 2016



**Inaugural
meeting**

UN NGO Committee for Rare Diseases

