

# NGO Committee for Rare Diseases (UN, NY)

ICORD Conference 2016  
Cape Town, October 21<sup>st</sup>

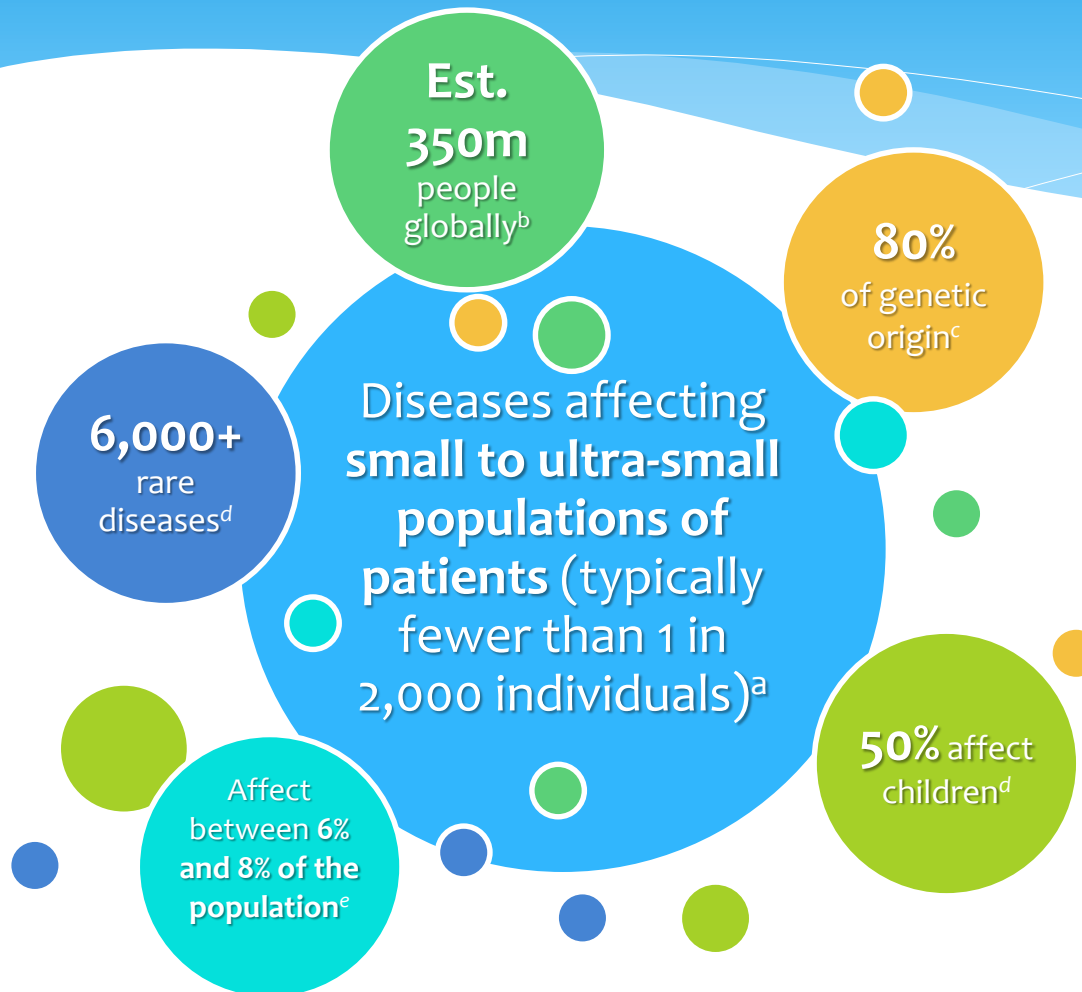


NGO COMMITTEE FOR  
**RARE DISEASES**

Anders Olauson  
**Chairman Agrenska Foundation**



# What are rare diseases?



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

<sup>b</sup> Estimate based on statistics available across different populations.

<sup>c</sup> [http://www.who.int/medicines/areas/priority\\_medicines/BP6\\_19Rare.pdf](http://www.who.int/medicines/areas/priority_medicines/BP6_19Rare.pdf)

<sup>d</sup> [http://www.orpha.net/consor/cgi-bin/Education\\_AboutRareDiseases.php?lng=EN](http://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN)

<sup>e</sup> 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)

# Common Challenges



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# The challenges of rare diseases

Although each disease itself is rare, the **challenges** facing people living with rare diseases are **common across diseases, and across borders**:

- \* Patients and experts are few and geographically scattered
- \* Little information about the diseases, knowledge and good practices are limited
- \* Little and fragmented scientific research as funding and resources are scarce
- \* Slow, or often inaccurate diagnosis
- \* Isolation: for patients, families, their health and social care professionals, their researchers
- \* Only 5% of rare diseases have adequate treatments

Rare

- \* 80% of rare diseases are genetic

Highly Complex

- \* Limit life expectancy
- \* Cause severe and complex impairments and disability
- \* No, or little, autonomy for the patients and their families
- \* Profound social and economic consequences

Highly Disabling

Serious unmet health and social needs affect individual's - and their families' - well-being, autonomy and fundamental human rights



\* “To understand how it is to have a child with a rare disease, you have to have a child with a rare disease yourself”

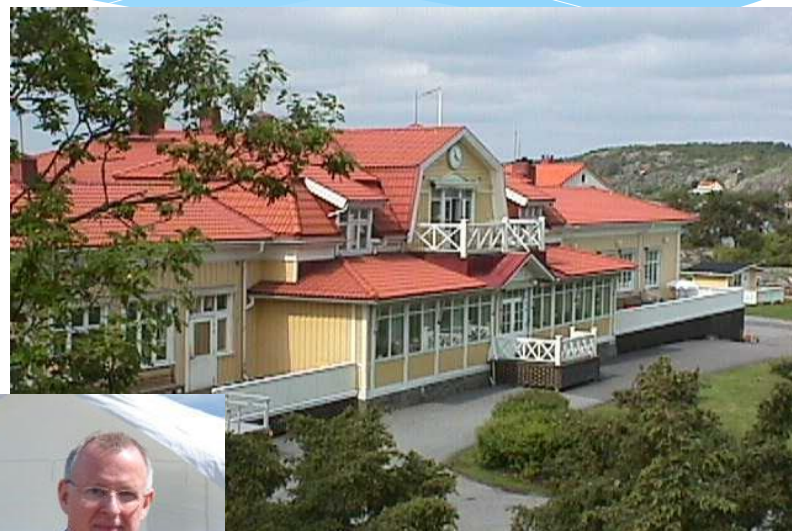
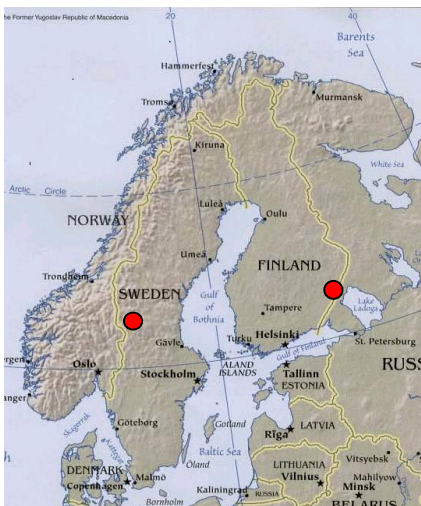
**Åke Martinsson, Sweden**



Roger,  
My teacher!



# Agrenska – a centre for people with disabilities, their families and professionals

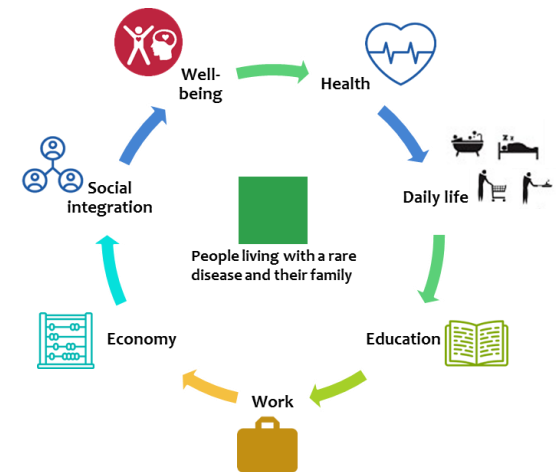


Queen Silvia Patron of Agrenska Sweden  
Mrs. Ilves, Patron of Eesti Agrenska

Today's Ågrenska started by combine the following parties;



- \* Hospital,
- \* School,
- \* Social,
- \* Patients
- \* the Agrenska Foundation





# Agrenska programs 1989 - 2016

- \* **Family program** – focus on rare diseases, since 1989 >5 000 families
- \* **Adult program** – focus on rare diseases
- \* **Respite care** - all kinds of disabilities
- \* **Staff with disabilities** – day centre
- \* **Personal assistance**
- \* **ADHD** consultant
- \* **Courses for professionals**
- \* **Research**
- \* **Conferences**
- \* **Projects/development**

# Why patients view, -Some reflections after more than 26 year's of experience



1. Too few professionals have knowledge of rare diseases and their impact on individual and family
2. Efforts incomplete or wrong
3. Long time before diagnosis set - needless suffering
4. Charged to the wrong parts of the system
5. Society's efforts are not coordinated - system competence is lacking in many of society - must be a carrier of knowledge
6. Need to discuss opportunities instead of obstacles
7. Must always explain and "defend" their difficulties / disability due to rarity and ambient ignorance
8. Need different stages of life to gain knowledge and meet others in the same situation

# Experiences worldwide today

- \* 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.

# Common Rights:

A life of dignity  
for all



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# Common Solutions



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# 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 marginalized, 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 need for **common solutions**

Increased collaboration can lead to **more data, more research, more treatments, greater awareness** and an improved, **global understanding** of rare diseases and good care practices.



Reduced inequalities and isolation for people living with rare diseases and their families at every stage of economic development.

# 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?



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# Why the United Nations?



World Health  
Organization



**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.**



# 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



# 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

# What will the Committee do?

Collect and share up-to-date information and research on the global dimensions of rare diseases

Develop and present reports, recommendations for actions and positions on rare diseases and associated issues

Host dedicated events to share information and to raise the profile of people living with rare diseases worldwide

Highlight current or potential opportunities for **improved and more integrated collaborations between all stakeholders**

**Create synergies** with other CoNGO committees and with NGOs in ECOSOC

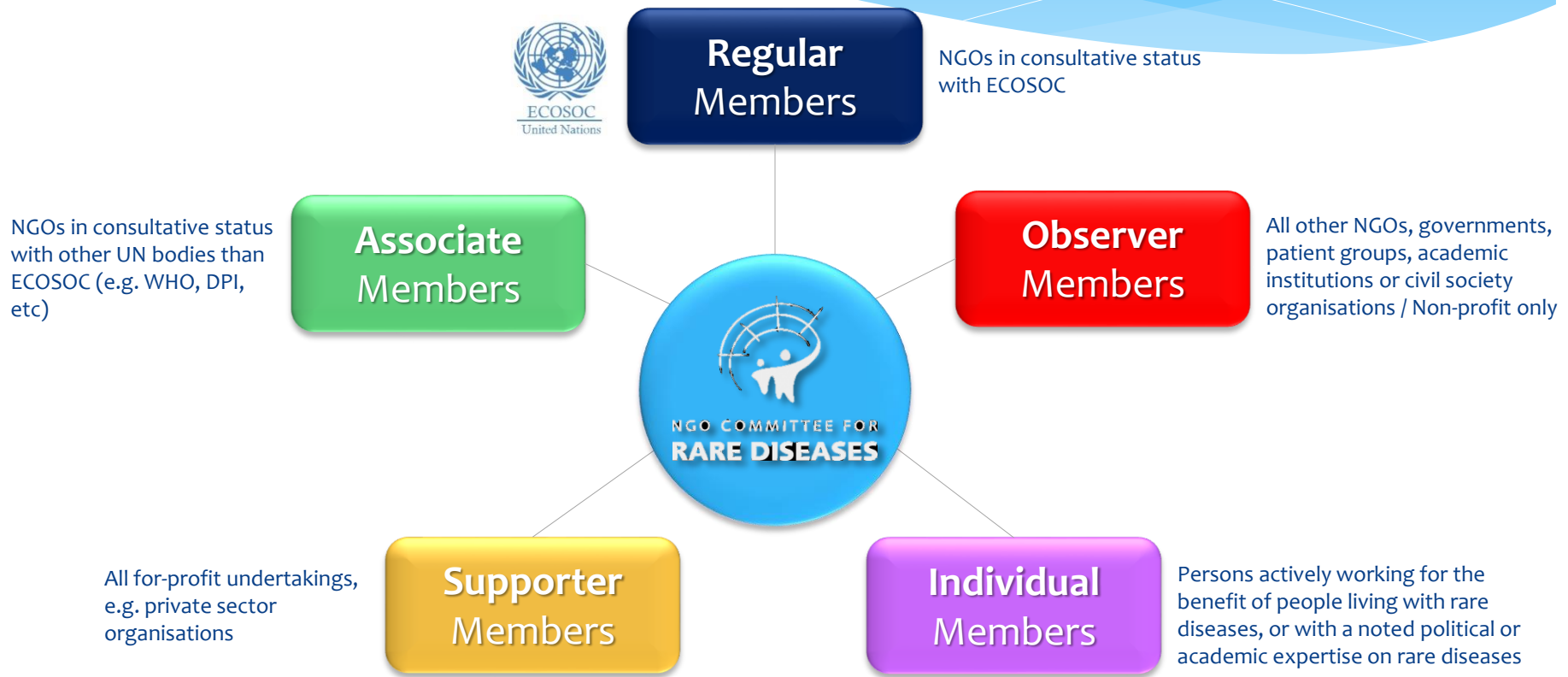
Encourage all relevant UN bodies to **better reflect aspects related to rare diseases into their mission and actions**



# 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*



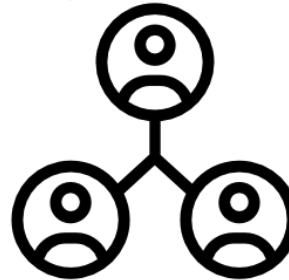
**Accepted  
as a CoNGO  
Committee**

*March 2016*



**By-laws  
approved by  
CoNGO  
president**

*April 2016*



**Establishing  
contacts  
within UN  
and  
member  
states**

*November 2016*



**Inaugural  
meeting**

# Why get involved in the Committee?

Although the primary purpose of the Committee is to bring together the international community of NGOs, the Committee is inclusive and requires contributions from all stakeholders.

## NGOs

- Strengthen your voice as part of a pioneering, global community for rare diseases
- Initiate dialogue with all rare disease stakeholders
- Bring your expertise to the discussion

## National Governments

- Engage with rare diseases experts, especially patients from your country
- Access up-to-date information about rare diseases collected globally
- Share experiences of rare diseases strategies to ensure no citizens are left behind
- Promote your country's voice, experience and noteworthy initiatives in a global dialogue

## United Nations Representatives

- Contribute actively to the post-2015 sustainable development agenda in catering to the unmet needs of some of the most vulnerable in society
- Create partnerships with the most influential rare diseases experts
- Raise the profile of rare diseases on the global agenda: make the invisible, visible

# SAVE THE DATE!



## Global Gathering for Rare Diseases: Inaugurating the NGO Committee for Rare Diseases

**Friday 11 November 2016**

Conference Room 8 (GA-1B-CR08), United Nations  
New York, NY 10017, USA

Under the patronage of:

**Queen Silvia of Sweden**

and

**The Conference of NGOs in Consultative Relationship with the United Nations (CoNGO)**

With the support of:

The Swedish Ministry of Health and Social Affairs

The Permanent Mission of Estonia to the UN

The Government of Malta

The French Minister of Social Affairs and Health (tbc)

The French Minister of Foreign Affairs and International Development (tbc)

**Watch the live webcast (link coming soon!)**

For more info: [contact@ngocommitteerarediseases.org](mailto:contact@ngocommitteerarediseases.org)



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