DIAGNOSING UNDIAGNOSED RARE DISEASE PATIENTS

Tools and resources to strengthen the voice of the rare undiagnosed community

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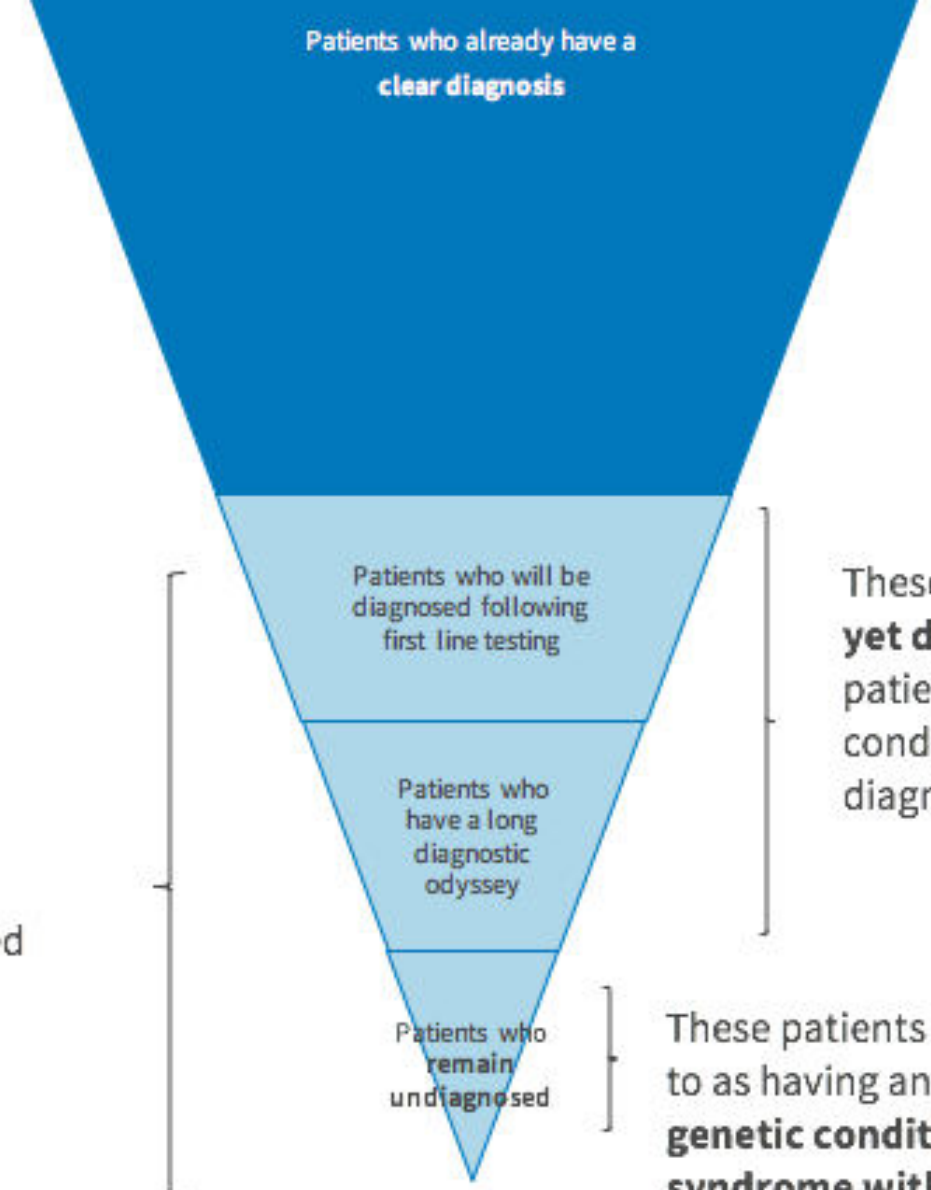
1st International Rare Disease Conference, 1 March 2021

AIMS OF SWAN EUROPE

- To provide a forum for sharing information.
- To work together to increase the visibility of syndromes without a name and/or undiagnosed genetic conditions and support the growth of the SWAN/undiagnosed support community.
- To facilitate networking by providing a point of contact for stakeholder engagement across Europe and supporting relationships with rare disease networks.
- To empower families affected by a syndrome without a name/undiagnosed genetic conditions.



These are patients who are currently **without a clear diagnosis**. Some of these patients will be not yet diagnosed and some will remain. undiagnosed.

These are patients who are **not yet diagnosed**. Some of these patients may have a rare condition, particularly if their diagnostic journey is long.

These patients are often referred to as having an **undiagnosed genetic condition** or a **syndrome without a name**. The cause of these conditions has not yet been understood.

CHALLENGES FACED

- Lack of public awareness.
- Social isolation.
- Access to testing/length of time for results.
- Lack of/poor quality information/care coordination.
- Impact of genome sequencing on clinical settings.

SOLVE RD COMMUNITY ENGAGEMENT TASK FORCE

- To provide a forum for sharing information.
- Support the interaction and engagement of Solve-RD and its stakeholders with different initiatives and networks existing in the field of diagnosis at the European and International levels.
- Ensure a transparent and patient-centred coordination of outputs through regular communication with ePAG representatives and the undiagnosed community.
- Ensure discussions and topics reflect the opinion and needs of the undiagnosed patient community.

SWAN UK
syndromes without a name

GENETIC ALLIANCE UK

Patient Journey through diagnosis

> *"It's a waiting game, but you tell a mum to wait when she's waited 15 years. It's difficult. – Nuria*

> *"People began to ask which side of the family it came from...It was a difficult time for us as parents. – Alexa*

> *"A diagnosis may be bad news, it may be very bad news or it may be no news. But all of that's OK and there's help and support for whatever spectrum you end up on. – Peter*

> *"We went around, travelling across the entire city to find a nursery for our son. It was impossible to have him accepted. – Gaston*

Information and training for patient organizations
Helplines
Information on secondary findings
Next Steps Toolkit
EUROGENTEST Guidelines
Experience Based Co-Design
RareConnect .org
ENSERIO study. Time to Diagnosis
Undiagnosed Photo Project
Protocol to support ultra-rare diagnosis
Training for professionals

EURORDIS
SolveRD
CHEO
European Patient Advocacy Group
EuroGentest
GLOBAL COMMISSION
UNIAMO
ISS
feder
SWAN UK
UDNI