

SWAN EUROPE

Keeping patients at the heart
of diagnostic advancements

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Patients who already have a
clear diagnosis

Patients who will be
diagnosed following
first line testing

Patients who
have a long
diagnostic
odyssey

Patients who
remain
undiagnosed

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.

PROMISE OF GENOMICS

ITALY: First national genome sequencing programme.

SPAIN: SpainUDP (undiagnosed rare diseases programme), part of Undiagnosed Diseases Network International (UDNP).

FRANCE: 2020 France – national genome sequencing programme.

UK: 100,000 Genomes Project → Genomic Medicine Service.



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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.



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AIMS OF SWAN EUROPE

1. To provide a forum for sharing information.
2. 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.
3. To facilitate networking by providing a point of contact for stake holder engagement across Europe and supporting relationships with rare disease networks.
4. To empower families affected by a syndrome without a name.



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SOLVE RD

COMMUNITY ENGAGEMENT TASK FORCE:

1. 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.
2. Ensure a transparent and patient-centred coordination of outputs through regular communication with ePAG representatives and the undiagnosed community.
3. Ensure discussions and topics reflect the opinion and needs of the undiagnosed patient community.



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THANK YOU



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