Patient organisations unite to support the 65,000 children born with an undiagnosed genetic condition each year across Europe

Friday 28 April 2017

On Undiagnosed Children's Day 2017, patient groups from all over Europe have come together to launch a groundbreaking coalition, SWAN Europe, uniting European and national patient organisations working to support and empower families affected by syndromes without a name.

More than half of all children taking a genetic test in Europe right now will never get a confirmed diagnosis for conditions that are often complex, life limiting and potentially life threatening.

Patient groups from all over Europe have come together to launch a groundbreaking coalition, SWAN Europe, uniting European and national patient organisations working to support and empower families affected by syndromes without a name.

More than 65,000 children are born in Europe each year with a syndrome without a name – a genetic condition so rare that it is often impossible to diagnose. Having no diagnosis leaves families in limbo. Children with syndromes without a name are impossible to track in European healthcare systems. Governments do not keep records of people who have not been given a confirmed diagnosis. This means they can have difficulties accessing appropriate health care and support.

This group of families is distinct from others affected by rare and genetic conditions and to access the support that families require, their specific needs must be acknowledged. We are working together, in a network of growing patients organisations to raise awareness and break the isolation of thousands of families in this situation. Member organisations of SWAN Europe support the International Joint Recommendations to

address specific needs of undiagnosed rare disease patients and bring recognition to this group as a distinct community.

Families with undiagnosed children experience strong feelings of isolation and exclusion as they often think that they are the only ones in this situation. They don't know that support is available. We want to change this.

Virginie Bros-Facer, Research Infrastructure Project Manager at EURORDIS-Rare Diseases Europe, a member of SWAN Europe, commented, "EURORDIS is a proud member of SWAN Europe. In our work, we advocate for the needs of all undiagnosed patients as well as the millions of patients who have already been diagnosed with a rare disease. In many European countries, even though rare diseases are a recognised public health priority, undiagnosed children remain one of the highly vulnerable populations whose specific needs have not been fully identified or assessed by national health and social authorities."

Ends.

Notes for editors:

Patient Case Studies
For patient case studies please contact the relevant national member organisation (listed below). For general enquiries please contact lauren.roberts@undiagnosed.org.uk.

Undiagnosed Children's Day
Undiagnosed Children's Day in Europe occurs on the last Friday of April each year, this year it falls on 28th April 2017. Similar national awareness days exist in USA and Australia although the date differs.

SWAN Europe
SWAN Europe is a coalition of groups, organisations and support networks working with families and/or patients affected by syndromes without a name and/or undiagnosed genetic conditions.

The aims of SWAN Europe are to empower families affected by syndromes without a name and:

- To provide a forum for sharing information.
- To work together to increase the visibility of syndromes without a name and/or undiagnosed genetic conditions.
- To facilitate networking by Providing a point of contact for stakeholder engagement across Europe, supporting relationships with rare disease networks and supporting the growth of the SWAN / Undiagnosed support community.
- To support the development of new support networks/organisations focused on syndromes without a name and/or undiagnosed genetic conditions.

- To work towards building a platform for the undiagnosed community to have a voice in policy development in Europe.

The current members of SWAN Europe are:

AnDDI-Rares
AnDDI-Rares is the French reference network for rare developmental abnormalities and intellectual disabilities. http://www.anddi-rares.org

Association Sans Diagnostic et Unique (ASDU)
ASDU is the French association that aims to provide technical and moral support for families confronted with developmental abnormalities and malformation syndromes without a diagnosis. http://www.asdu.fr

EURORDIS-Rare Diseases Europe
EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 700 rare disease patient organisations from more than 60 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting patients, families and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services. Follow @eurordis or see the EURORDIS Facebook page. For more information, visit www.eurordis.org

FEDER
http://www.enfermedades-raras.org/

Hopen
http://fondazionehopen.org/home

Objetivo Diagnostico
www.facebook.com/objetivodiagnostico/

Platform ZON
http://www.ziekteonbekend.nl/

SWAN UK (syndromes without a name) - United Kingdom
SWAN UK (syndromes without a name) is the only dedicated support network for families affected by undiagnosed genetic conditions in the UK. It is run by the charity Genetic Alliance UK. www.undiagnosed.org.uk. Follow @SWAN_UK on Twitter, find us on Facebook/SWANchildrenUK or follow us on Instagram, SWANchildrenUK.
Genetic Alliance UK is an UK national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 190 patient organisations. www.geneticalliance.org.uk

UNIAMO
UNIAMO is the Italian federation for Rare Diseases.
http://www.uniamo.org/

Prospective members should contact info@SWANeurope.eu.

International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients

The International Joint Recommendations to Address Specific Needs of Undiagnosed Rare Disease Patients was published in October 2016 on behalf of patients living with undiagnosed and rare diseases across Europe, North America, Australia and Japan.

SWAN UK (the support network run by Genetic Alliance UK), the Wilhelm Foundation, EURORDIS-Rare Diseases Europe, Rare Voices Australia (RVA), the Canadian Organization for Rare Disorders (CORD), the Advocacy Service for Rare and Intractable Diseases' stakeholders in Japan (ASrid) and the National Organization for Rare Disorders (NORD) jointly submitted a list of recommendations to address the specific needs of patients without a diagnosis.

The recommendations can be found here:
http://download.eurordis.org.s3.amazonaws.com/documents/pdf/Undiagnosed-International-Joint-Recommendations.pdf