4th
Scientific International Congress on
Spinal Muscular Atrophy

GHENT

14th — 16th March 2024

Organised by

SMA EUROPE

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ABSTRACT BOOK

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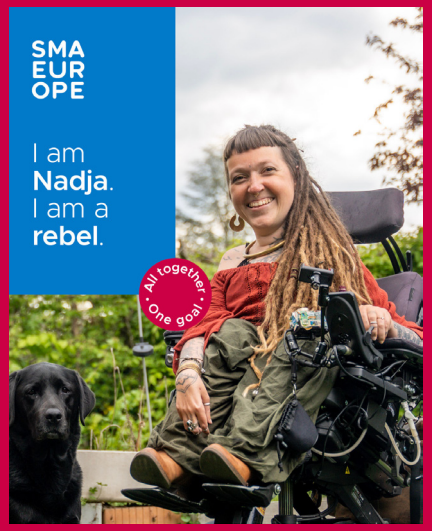
#WeAreUnique
2023 Awareness Campaign Vernissage

SMA has many different faces.

Our SMA Europe community is an example of how diverse the condition is, and how many different manners exist when it comes to living with it.

Traveler, Explorer, Journalist, Rebel… eleven individual stories which, together, resonate in one powerful poem that we want to share with you.



Join us and visit the Vernissage:
ICC Ghent, 1st floor, next to the Auditorium

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Welcome

SMA Europe takes great pride in inviting you to the 4th International Scientific Congress on Spinal Muscular Atrophy (SMA), from 14th to 16th March 2024 in Ghent, Belgium. Our Congress is the largest scientific congress dedicated to SMA and brings together scientists and young researchers as well as clinicians and other health-care professionals from all over the world.

SMA Europe is a non-profit umbrella organisation of SMA patient organisations from across Europe. We work together to create a better world for all those living with SMA. One of our core activities is to foster patient-relevant research in the field of SMA, to communicate the value generated from research, and, consequently, to ensure future support for research within our community.

The goal of our scientific congress is to bring together an international and multidisciplinary group of scientists and health-care professionals. We provide a venue to present and exchange breakthrough ideas relating to SMA, especially also in light of the patientrelevance of their findings, and to cement existing and stimulate new collaborations.

Equally, our congress is a platform for talented young researchers. Together with experienced scientists, health-care professionals, and patient experts, they will discuss, debate and dissect significant new developments and advancements related to SMA. Moreover, they will share their visions for the future of SMA research by providing an opportunity to exchange scientific evidence and clinical experiences. Therefore, we trust that the conference will lead to ever-improved treatment and care for patients living with SMA.

The International Scientific Congress on Spinal Muscular Atrophy is a unique occasion to meet face-to-face with colleagues who are as passionate about advancing the field of SMA as you are.

All together. One Goal.

SMA Europe very much looks forward to welcoming you in Ghent!

Nicole Gusset
SMA Europe CEO and President

SMA EUROPE

About us

SMA Europe is a non-profit umbrella organisation of spinal muscular atrophy (SMA) patient organisations from across Europe. We work to bring effective treatments and optimal care to everyone living with SMA.

Together, through greater understanding, we will create a better world for all those living with SMA.

All together. One goal.

Our priorities

Research

Our mission is to be active and progressive in the search for treatments for SMA. We do this through promoting and generating patient-relevant data. We are supported by a Scientific Advisory Board (SAB), composed of neuroscientists and neurologists with particular expertise in SMA.

Through our research programme, we:

- Seek to set patient-relevant research priorities
- Promote these patient-relevant research priorities in our Call for Research Proposals and our scientific congresses
- Systematically research and assess the needs and wants of people living with SMA
- Identify data gaps that are relevant to patients and fill those by:
    - producing our own patient-relevant research projects and publishing the outcome in peer-reviewed journal
    - stimulating, (collaboratively) supporting and funding research that addresses these gaps
    - facilitating communication between stakeholders in this field.

We also build our members' capacity to understand the relevance and processes of research, to allow them to become partners in funding research and in meaningfully contributing to discussions and solutions. In so doing, we make sure SMA research delivers on patients' unmet needs from a clinical, care and quality of life perspective.

We do this because we believe that developing a treatment that can truly help improve the lives of people living with SMA should be rooted in a firm understanding of the challenges those people face in their daily lives, their needs and the trade-offs they are willing to make to gain relief.

To ensure the creation of valuable treatments, all aspects of the health care system, including research prioritisation, product development, trial design, regulatory approval, access, reimbursement and treatment decisions, will need to align with their needs.

Therapy and Care

SMA Europe strives to accelerate progress in the diagnosis, treatment and care of people with SMA.

To this aim, we engage in dialogue with all relevant stakeholders, to ensure the needs and wants of people living with SMA across Europe are taken into account during the entire drug development process.

To justify a seat at all relevant tables and to be able to provide meaningful, qualified and evidence-based input, SMA Europe continuously educates and prepares individual patient advocates in key knowledge areas. In parallel, SMA Europe strives to collaborate with all key stakeholders as a respected partner, especially in the areas of drug development and regulatory affairs.

Healthcare Systems, Policy and Access

Access to diagnosis, treatment and care is fragmented in Europe. SMA Europe strives for unrestricted access to optimal available medicine, treatment, care and diagnostics, regardless of location, age, mobility or SMA type. This is the only outcome which will end the access inequalities that SMA families continue to live with today.

We address this issue by mapping and centralising information around access throughout Europe. We identify data gaps that influence access and we promote research to fill them. We support our members by sharing knowledge and coaching them to advocate efficiently in their own country. At a European level, we partner with and influence all stakeholders in relevant areas of healthcare and research, wherever impact can be made, bearing in mind that responsibilities in access tend to fall at national level and are limited at European level.

SMA EUROPE
Member organisations

Belgium
SMA Belgium
www.spierziektenvlaanderen.be
www.telethon.be

SMA Belgium is an umbrella organisation created by the SMA working groups of Spierziekten Vlaanderen (Flemish neuromuscular organisation) and ABMM (Walloon neuromuscular organisation).

SMA Belgium is dedicated to building communication networks between families with SMA about progress in scientific research, available treatment and public assistance.
Finally, but not least, to strengthen the patient's voice in the drug development process, to collaborate with various stakeholders to optimise the drug-development path from the laboratory to the patient.

Cyprus
MDA Cyprus
www.mdacyprus.org

The Cyprus Muscular Dystrophy Association (MDA Cyprus), was founded back in 1986 and its members are children and adults from all over Cyprus. MDA Cyprus consists of patients who suffer from neuromuscular diseases such as Duchenne Muscular Dystrophy, Myasthenia Gravis, Amyotrophic Lateral Sclerosis (ALS), Peripheral Nerve Diseases like Charcot-Marie-Tooth Disease, Spinal Muscular Atrophy and more than 50 other types of Muscular Dystrophy.

Czech Republic
SMÁci
www.smaci.cz

The Czech SMA Patient Organisation – SMÁci, supports patients and their families with their SMA quest, striving to establish a communication channel between all parties involved and helping to achieve a smooth execution of all available steps to improve quality of life. Since its creation in 2016, SMÁci has had a key role in advocating for access to treatment for people with SMA in Czech Republic.

Denmark
Muskelsvindfonden
www.muskelsvindfonden.dk

The Danish Muscular Dystrophy Foundation unites people and relatives with all types of neuro muscular diseases. The organisation consists of four units: a member based association; an event and fundraising organisation which organises a music festival and a circus-theatre; a highly specialised centre for neuromuscular diseases; and the Musholm holiday, sports and conference centre, internationally awarded for its outstanding accessibility and run as a social enterprise. Together, these four units work on advocacy, awareness, research, and empowerment to improve treatments and create opportunities for living a good life with a neuro muscular disease.

Finland
SMA Finland
www.smafinland.fi

SMA Finland was founded in 2018 by group of active SMA patients and parents with children who have SMA. The primary goal of the association is to provide access to medical treatment to all SMA patients, despite the type or age.
Other important goals for SMA Finland are also to improve the quality of life of individuals with SMA and to create awareness about SMA with authorities, healthcare professionals and the general public.

France
AFM-Téléthon
www.afm-telethon.fr

The French Muscular Dystrophy Association (AFM) federates patients with neuromuscular diseases and their parents. Thanks in great part to donations from France's annual Telethon, the AFM-Telethon has become a major player in biomedical research for rare diseases in France and worldwide. It currently funds about 37 clinical trials in different genetic diseases affecting the eye, blood, brain, immune system, and muscles... Thanks to its Genethon research lab, the AFM-Telethon stands out through its unique ability to produce and test its own gene-based medicines.

Germany
DGM
www.dgm.org

The German Society for people with Muscular Diseases (DGM), founded in 1965 at the initiative of parents, is the oldest and largest patient organisation for people with muscular diseases in Germany. Its main objective is to the advance research in the field of muscle disorders and treatments.

DGM provides advice, assistance, and the opportunity to exchange experiences with other stakeholders, also being committed to the concerns of the other stakeholders within health policy.

Greece
MDA
www.mdahellas.gr

MDA (**Muscular Dystrophy Association Hellas**) focuses on improving the lives of individuals with neuromuscular disorders by working with government agencies, universities and various institutions and individuals to further research on neuromuscular disorders and to promote early diagnosis. The organisation runs events and education programmes for patients focused on advocacy, clinical trials, novel medicines, and technologies while also supporting and educating doctors. MDA Hellas has established and supports 3 Specialised Neuromuscular Units in Greece.

Hungary
SMA Hungary
www.smahun.hu

The primary goal of the Foundation is not to directly support patients, but to support domestic treatment (e.g., additional personal resources), to exchange information about treatment options and to organise and finance events related to this (in particular study trips abroad, lectures of foreign doctors in Hungary).

Iceland
FSMA Iceland
www.fsma.is

FSMA Iceland was formally established in 2002 and is an association of families and individuals who suffer from Spinal Muscular Atrophy (SMA). Its purpose is to protect the interests of the persons with SMA and their families and to contribute to finding a cure. FSMA does this by fundraising to support research into the science of the disease; providing information to members of FSMA in Iceland on the progress of research on the disease, as well as other useful information related to the disease; to hold meeting on issues SMA; to disseminate information in the media and to the media about issues pertaining to SMA; to contribute to the transportation of individuals with SMA and their families.

Ireland
SMA Ireland
www.smaireland.com

The Spinal Muscular Atrophy Ireland Foundation is the collective voice for the adults and children with SMA in Ireland. The organisation supports people and families by: providing information and a network of contacts; raising awareness of SMA and campaign for screening; lobbying Government and the Health Service for access to treatment; liaising between pharmaceutical companies and patients/families; facilitating ongoing research by coordinating patient volunteers; and maintaining connections with SMA organisations in other jurisdictions.

Italy
Famiglie SMA
www.famigliesma.org

The Association of Families of SMA is a non-profit NGO founded in 2001 by a group of parents of children with SMA. The Association is a point of reference for medical and scientific researchers and for all the families of children with SMA. It aims to inform families about progress in scientific research, available treatments and assistance to which they are entitled to from public institutions; to promote and support scientific research in SMA and possible therapies; to communicate developments in clinical trials of drugs and therapies as well as the participation of Italian research organisations in clinical trials for SMA abroad, also solving legal problems, bureaucratic and organisational related to such participation.

Macedonia
Stop SMA
www.stopsma.mk

The Association of persons with Spinal Muscular Atrophy STOP SMA is an association of citizens, established for the purpose of realising, protecting, and promoting the rights and interests of the persons with SMA and their custodians. This includes their rights and interests regarding health protection as well as their rights to treatment, social protection, education, and employment.

The Netherlands
Prinses Beatrix Spierfonds
www.prinsesbeatrixspierfonds.nl

The Prinses Beatrix Spierfonds, a foundation for over 200.000 people with a neuromuscular disease in the Netherlands, aims to eliminate all neuromuscular diseases by means of scientific research. The organisation finances and stimulates research

aimed at developing therapies for neuromuscular diseases. Because this can be a long and strenuous process, the organisation simultaneously supports research in improving quality of life.

The Netherlands
VSN - Spierziekten Nederland
www.spierziekten.nl

Spierziekten is an association of and for people with a neuromuscular disease. Its activities consist of providing information, organising mutual support, and stimulating scientific research, including international cooperation in the fields of research and of the development of therapies.
Spierziekten runs a series of initiatives focused on the improvement of social and medical care for people with neuromuscular diseases and organises various information and dissemination activities each year.

The association collaborates closely with relevant experts to improve diagnostic procedures, care, rehabilitation, and genetic counselling while also maintaining a network of regional groups and national diagnosis-bound support groups.

Poland
Fundacja SMA
www.fsma.pl

The Polish SMA foundation, formed by parents of children with SMA, has the following goals: to conduct activities for people with SMA and their loved ones, aimed at combating exclusion, increasing independence and improving their quality of life; to increase the awareness of SMA, by disseminating knowledge in genetics, diagnostics, standards of care and treatment methods; to increase the availability of methods and techniques for diagnostic, therapeutic, rehabilitative and related products and technology solutions; to support system solutions, particularly in health care and social security, taking into account the needs of people with SMA and their loved ones.

Portugal
APN
www.apn.pt

APN (**Associação Portuguesa de Neuromusculares**) focuses on creating and promoting better quality of life for people living with neuromuscular diseases. Its members are people with muscular diseases, family members, doctors, and other health professionals. APN's actions include advocacy, direct support to people with neuromuscular diseases and their members and support to medical research.

Romania
Asociatia SMACARE
www.amiotrofie-spinala.ro

SMACARE Association is a non-governmental and non-profit organisation that aims to protect the rights and interests of people affected by spinal muscular atrophy and to improve their lives. The association was founded at the initiative of some parents whose daughter is affected by Spinal Muscular Atrophy Type II. Helped by relatives and friends and encouraged by doctors decided to start on this road to change the mentality of Romanians about people with disabilities, to make this disease known and to build a communication network between people with muscular spinal atrophy, their families and doctors.
Asociatia SMACARE aims to:

- Improve the quality of live of individuals with Spinal Muscular Atrophy;
- Raise awareness about Spinal Muscular Atrophy with the general public, health-care providers, governmental organisations;
- Build a community for Spinal Muscular Atrophy families and individuals;
- Funding for Spinal Muscular Atrophy projects;

Russia
SMA Family Foundation Russia
www.f-sma.ru

The SMA Family Foundation Russia, established by parents of children with SMA, supports and empowers families with people living with SMA. The Foundation focuses on advocating for better services, raising public awareness and funding family's special needs that are not reimbursed by national healthcare service. Its main goals are: to build strong basis for the improvement of the quality of life of individuals with SMA and their families; to fulfil the strong need of information and best care practices; and to promote the development of medical and non-medical care for SMA.

Serbia
SMA Serbia
www.smasrbija.rs/en/support

SMA Serbia, established by parents of children with SMA and adults living with SMA, aims at increasing the quality of life of those living with SMA and their families. The association strives to raise public awareness, to protect the interests and the rights of people with SMA and their families and to improve their social care and medical support. The aim is to build a community for SMA families and individuals, to advise them and to make their lives easier. SMA Serbia's final intention is to get the right treatment for every single patient, children and adults, so that they grow, improve and prosper in many fields in life.

Spain
FundAME
www.fundame.net

Spinal Muscular Atrophy Foundation (FUNDAME) is a non-profit, private foundation, established in 2005 and made up of patients affected by SMA and their relatives. FUNDAME strives to find ways to improve the quality of life of those affected by SMA and to promote research into this disease.
FUNDAME supports research at both national and international level, in order to bridge the gap between today and the day a cure for SMA is available. In the meantime, FundAME seeks ways to improve the quality of life of those affected by the disease.

Sweden
NSMA
www.nsma.nu

NSMA (Nätverket för spinal muskelatrofi) is a patient association which aims to bring together people who work and live with SMA in order to exchange, assist and inform; to be the natural platform for exchange and support for people living or working with SMA in Sweden; to raise awareness about SMA in Sweden; to influence public opinion and policy makers in different social and health care organisations on issues related to SMA; and to establish a Swedish care program for SMA with information and guidelines for different treatments.

Switzerland
SMA Schweiz
www.sma-schweiz.ch

SMA Schweiz operates to develop and optimise therapies for people with SMA. The goals of SMA Schweiz are to strengthen the patients' voice in the drug development processes, to collaborate with various stakeholders to optimise the drug-development path from the laboratory to the patient, and to educate people affected by SMA and their families, as well as the general public.

Turkey
SMA Benimle Yürü
www.smabenimleyuru.org.tr
www.sma.org.tr

In Turkey, the SMA Community, is being represented by delegates from two organisations: SMA Benimle Yürü and Turkey SMA Foundation.

Ukraine
CSMA
www.csma.org.ua

CSMA (Children with SMA) unites both parents of children and people with SMA to foster exchanges of ideas to solve, at least partially, existing problems. CSMA also supports the maintenance of a national registry of patients with SMA and provides information on SMA care related topics.

United Kingdom
SMA UK
www.smauk.org.uk

SMA UK was established by a mother whose baby died aged 7 months from SMA. She set up the charity under the name of 'Jennifer Trust for Spinal Muscular Atrophy', now SMA UK, to offer support and hope to other families affected by the different types of SMA. SMA UK advocates for better services and access to new treatments so that people affected by SMA are supported, empowered and enabled to live full lives. The charity is also committed to help fund and facilitate research and to raise public and professional awareness of SMA.

Newborn Screening

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Six months of newborn screening for spinal muscular atrophy in Croatia

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Spinal muscular atrophy (SMA) is a neuromuscular and neurodegenerative disease caused by homozygous deletion of exon 7 of the *SMN1* gene in 95 % of cases. Until recently, SMA was the most common genetic cause of death at an early age. However, with the development of therapies that modify the natural course of the disease, the prognosis of patients with SMA has significantly improved. All three existing therapies, nusinersen, onasemnogene abeparvovec and risdiplam, are available in the Republic of Croatia and applied according to the indications of the Croatian Health Insurance Fund. The best outcomes in treatment are achieved if the therapy is applied before the symptoms of the disease appear, which is why newborn screening (NBS) for SMA is of key importance in the treatment process. It is expected that a certain number of patients detected by NBS will already be symptomatic at the time of diagnosis. A small number of patients, around 5 %, cannot be detected by NBS.

Herein, we present the results of the SMA NBS during the first 6 months of the pilot project in Croatia and verify the suitability of the Targeted qPCR SMA assay (ZenTech, Belgium) for SMA NBS.

The first-tier test for SMA NBS developed by ZenTech is based on quantitative polymerase chain reaction (qPCR) from a dried blood spot and detects homozygous deletion of exon 7 of the *SMN1* gene. The second-tier test is based on MLPA technique (MRC Holland, P021 kit).

The pilot project of SMA NBS in Croatia started on March 1st, 2023 in the Dept. of Laboratory Diagnostics of University Hospital Centre Zagreb. By September 1st, 2023, a total of 16 035 newborns were tested. 0.05 % of parents / guardians refused screeening. Two SMA patients were detected and their diagnosis was confirmed by MLPA analysis on the 11th and 12th day of life, respectively. One patient had 3 *SMN2* copies, and the other had 6 *SMN2* copies. There have been no false positives or false negatives to our knowledge so far. The incidence of SMA of 1 in 8 017 determined during the NBS pilot study in Croatia is consistent with previous studies of SMA prevalence in Croatia. SMA NBS allows the early diagnosis of SMA as well as timely application of therapy, which prevents disease progression. Our results indicate that the ZenTech assay can be reliably used in SMA NBS, as well as the importance of adding SMA in the national screening program of the Republic of Croatia.

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Revolutionizing Spinal Muscular Atrophy Prevention in Serbia: Implementing a Mandatory Statewide Newborn Screening

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Spinal muscular atrophy (SMA) is the prevalent genetic cause of childhood mortality. Pioneering treatments yield utmost advantages only within the presymptomatic phase, underlining the medical and ethical significance of newborn screening.

In 2022, the Centre for Human Molecular Genetics initiated a pilot study of the newborn screening for SMA, working closely alongside the University Children's Hospital Tirsova and Association SMA Serbia. The aim was to lay the foundation for the implementation of statewide newborn screening for SMA in Serbia by conducting screening for ~8000 infants from the Obstetrics and Gynaecology Clinic Narodni Front over the course of a year. In the subsequent year, we expanded the initiative to include another maternity hospital located outside Belgrade, introducing sample shipment via postal services and extending screening accessibility to a greater number of infants.

In the initial year, 6 950 newborns underwent testing, revealing SMA in two unrelated infants. Subsequently, an older sibling of the first newborn, although asymptomatic at the time, was also tested at the age of 16 months, and SMA diagnosis was confirmed in this child as well. All three children received therapeutic interventions in <1 month from birth. To date, they have exhibited no signs of SMA, and there have been no false-negative outcomes among the newborns who tested negative during the screening. In the second year, an additional 5 000 newborns underwent testing.
As frontrunners in this field in Serbia, we orchestrated harmonized efforts across various tiers of healthcare, established screening and diagnostic algorithms and follow-up protocols. Our extensive efforts were primarily aimed at elevating awareness among all stakeholders about the critical importance of early disease detection. In this transformative journey, we transitioned from being isolated individuals and visionaries who championed a singular idea to an entire community and nation that now acknowledges the paramount significance of newborn screening. As a result, a total of 11 950 infants underwent testing during the 17-month pilot project, culminating in the rapid incorporation of newborn screening for SMA into the national screening program, effective as of September 14th 2023.

Timely detection and treatment can transform SMA into a manageable condition, and there is substantial evidence supporting its inclusion in state-wide screening programs.

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