European Foundation for SATB2-Associated Syndrome (SATB2 Europe)

Evropska fundacija SATB2, ustanova za SATB2 sindrom

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SATB2 Europe's ANNUAL REPORT for the financial year 2024

(1.1.2024 - 31.12.2024)

I. BUSINESS REPORT

1. INTRODUCTION

1.1. Introduction of the Foundation

The Foundation is a non-profit, charitable organisation in the field of health and rare diseases. Its purpose is of general benefit and lasting, aimed at improving the quality of life of individuals with SATB2 syndrome and their families by enabling research for the discovery and development of targeted treatments and by providing better medical and rehabilitation care in the European Healthcare System.

1.2. Development of the Foundation

The idea for the Foundation was born in January 2021, during the first virtual meeting of three families who have a child with SATB2 syndrome. This was followed by the strategic plan and everything needed to set up the Foundation at the relevant Slovenian Ministry - Ministry of Health and the registration in the Register of Foundations on 21 July 2021. At the same time, we informed European families about our vision and had meetings with 18 European countries as well as the USA and Australia. We have created a website (temporary) and everything we need to start our business.

1.3. Leadership

The Board of Directors is composed of , namely Erika Stariha (Chairperson of the Board, Slovenia), Jenny Li Örsell (Board Member, Sweden), Paul Maksys (Board Member, Austria). We manage the Foundation in accordance with our internal rules. Currently, the President, with the help of a board member (Paul Maksys),

also takes care of the financial part, the administrative work and the communication with the target audience. All board members as well as other staff members work on a voluntary and unpaid basis.

1.4. Committees - Initiatives

The Foundation has 2 main initiatives, CareSATB2 and CureSATB2, the first with the aim of improving medical and rehabilitation care within the European Health System and the second with the aim of promoting and facilitating research to expand knowledge and treatment of SATB2 syndrome. The CareSATB2 initiative is led by Erika Stariha and CureSATB2 by Jenny Li Örsell. Within each initiative, individuals from different countries are involved and the team is still forming.

1.5 Overview of activities

The Foundation has four main areas of activity:

- Promoting and facilitating research into the understanding and treatment of SATB2 syndrome.

- Improving the rehabilitation, care and healthcare of individuals with SATB2 syndrome within the European healthcare system.

- Providing resources to help and support families who have children or adults with SATB2 syndrome.

- Raising awareness of SATB2 syndrome among the scientific, medical, rehabilitation, educational and general public.

Accordingly, we carry out the following activities:

- Organisation of professional events, meetings and other educational activities.

- Networking between researchers, doctors, parents and other organisations or companies in the field.

- Networking between doctors and clinics in Europe and promoting the establishment of reference centres for SATB2 syndrome patients and clinical guidelines for the diagnosis and treatment of SATB2 syndrome.

- Help and support families who have children or adults with SATB2 syndrome, in particular by raising awareness and providing information on activities and new developments in this field.

- Raising awareness of SATB2 syndrome among the scientific, medical, rehabilitation, educational and general public.

- Raising funds, in particular donations from individuals and legal entities, applying for grants to carry out the purpose of the Foundation.

1.6. Environmental impact

SATB2 syndrome is a rare neurodevelopmental disorder. Currently, there are about 600 diagnosed patients in the world (about half of them are from Europe). The facts described above show that it is essential to work internationally if we are to be successful in achieving our goals. In 2021, there were only 5 national associations in Europe (2 of which were founded in 2021 year), and the other countries have no formal associations, so we have set up the European Foundation to help those who do not have a national association and to be a strong partner in discussions with organisations outside Europe. In the year 2022 there were no new formal SATB2 related organisations in Europe, but we helped to form active groups of parents in Poland, Portugal and Greece to work on national level and to help us connect their clinicians with the international network.

2. PROGRAMME

2.1. Achieving the 2023 Plan

We have set realistic targets and have largely achieved them.

2.1.1. Core business

In the past year, we have regularly published news and created campaigns for Rare Disease Day and SATB2 Syndrome Awareness Day (Instagram, Facebook, LinkedIn, YouTube channel). We continued the renovation and upgrade of our website, which is still ongoing.

We actively participated in ERN ITHACA (Patient Council) meetings and attended the annual members' meeting in Bucharest. We remained founding members of the Genetic Alliance and members of Eurordis. Additionally, we became members of the Association of Patient Organizations of Slovenia, in the section for rare diseases. We also joined the "Patient and Citizen Pillar" at BBMRI-ERIC – an international biobank network.

We maintained contact with all SATB2 organisations and actively collaborated with them (USA, Australia, France, Italy, Spain, England, and the Netherlands & Belgium). Furthermore, we attended an international conference on a related neurodevelopmental syndrome – CTNNB1 syndrome, where we expanded our knowledge and networked with experts in this field.

2.1.2. CareSATB2 initiative

In April 2024, we attended the EURO Neurodevelopmental Disorders (EURO NDD) conference in Lisbon, where we successfully networked with clinicians and researchers, resulting in new connections with the potential for future projects, pending successful funding.

In June, our board president participated in the School of Scientific Innovation and Translational Medicine Research in Barcelona, organized by Eurordis – Open Academy School link. She also received partial reimbursement for travel expenses, as only five participants were selected. link

In December 2024, the board president attended the annual ERN ITHACA General Assembly and gave two active presentations: sharing good practices (guideline project) and sharing a personal story on the challenges of finding a causal diagnosis. link

Guidelines

In 2024, we continued one of our foundation's main projects: the preparation of the first global guidelines for the care and treatment of SATB2 syndrome. The project is methodologically supported by ERN ITHACA, while we independently established a network of interested clinicians worldwide based on information gathered from parents of children with this syndrome. This is a significant milestone for the field we represent, and achieving it has required substantial effort and advocacy.

Over the past year, with the help of parents in our consortium, we compiled a set of 48 clinical questions. Using a survey shared with the SATB2 community in eight languages, we gathered insights on the importance of each question, helping to define the content scope of the first guidelines.

European Reference Center for SATB2 Syndrome

We successfully negotiated the establishment of an Expert/Reference Center in the Netherlands, with a planned visit in 2025 for final confirmation and operational launch.

Awareness

With our social media posts (primarily on LinkedIn), we achieved a strong response and wide reach, contributing to raising awareness about the challenges of rare diseases both in Slovenia and beyond.

2.1.3. CureSATB2 initiative

Towards the end of 2024, we began formalizing the **Scientific Advisory Board** for our foundation. We launched new projects in collaboration with the **Faculty of Pharmacy in Ljubljana** and laid the groundwork for partnerships with a **laboratory in Madrid** and a **center in the Czech Republic** in the field of **translational science** for our syndrome.

II. FINANCIAL STATEMENTS

1. Notes to the financial statements

The basis on which the Annual Report of the Foundation is based is the Accounting Act. It refers to the Slovenian Accounting Standards (2016) , in particular to the Slovenian Accounting Standard 34 - Accounting solutions in non-profit organisations - legal entities governed by private law. The Uniform Chart of Accounts is also taken into account. In accordance with Article 11 of the Accounting Act, the financial statements and the report on the Foundation's operations are drawn up for the financial year, which is the same as the calendar year. The assets and liabilities are reported as at 31 December. The income, expenditure and surplus or deficit for the period from 1 January to 31 December are shown in the income and expenditure account and the financial statements include information for the current and the previous accounting period. The accounts

shall be kept in such a way as to permit the necessary information to be presented for the preparation of the annual balance sheet and the economic outturn account. The accounts shall be kept on the double-entry basis. As regards the separate keeping of accounts for profit-making and non-profit-making activities, revenue shall be recorded according to the type of activity and the proportion relating to profit-making activities shall be calculated. This calculated proportion is then used to separate the costs into a profit and a non-profit part. When submitting the annual report to the Agency of the Republic of Slovenia for Public Records and Services, the Instruction on the submission of annual and final reports and other data of business entities shall be followed. The deadline for submitting the annual report shall be the last day of February of the current year for the preceding calendar year.

2. Balance sheet

	Assets (EUR)	Liabilities (EUR)
Cash	2.637,12	
Founding input		1.200,00
Current year's surplus of income over expenditure		976,93
Previous years' surplus of income over expenditure		460,19
Total	2.637,12	2.637,12

3. Notes to the balance sheet

Cash is represented by the cash in the transaction account opened with Unicredit bank and the Pay pal account totalling EUR 2.637,12. The founding contribution amounts to EUR 1.200.

The balance sheet shows a surplus of EUR 976,93 of income over expenditure for the current year and a surplus of EUR 460,19 of revenue over expenditure for previous years**.**

4. Income statement

V EUR	2024	2023	2022
Revenue	1.103,10	150,01	445,80
Expenses	126,17	127,04	663,40
Surplus of revenues	976,93	22,97	
Surplus of expenses			217,60

5. Notes to the Income and Expenditure Account

Revenue and expenses are recognised on an accruals basis and are recorded in the accounting period to which they relate. Revenue includes income from donations of EUR 1.103,09 and interest income of EUR

0,01. The cost of goods, materials and services includes the cost of goods, services and the cost of payment and bank charges, amounting to EUR 101,17. As revenue exceeds expenditure, a revenue surplus for the period of EUR 976,93 has been identified. All revenue is non-profit making and therefore all expenses are also non-profit making.

III. CONCLUSION

The report was drawn up by the President of the Board and presented at the virtual Board meeting on 19.2.2025, where it was unanimously adopted.

The signatory is the official representative of the Foundation and the Board president, Erika Stariha.

At Šentjošt nad Horjulom, 24.2.2025

Erika Stariha