SATB2-ASSOCIATED SYNDROME

Awareness day August 22nd

FREQUENT CHARACTERISTICS

ABSENT OR LIMITED SPEECH

DENTAL ANOMALIES

BONE ANOMALIES

CLEFT PALATE

BEHAVIOURAL ISSUES

INTELLECTUAL DISABILITY

UNIQUE SMILE

DEVELOPMENTAL DELAY

FEEDING ISSUES

EPILEPSY

JOVIAL PERSONALITY

SLEEPING DIFFICULTIES

KEY FACTS

- **SATB2-Associated Syndrome (SAS)** is also known as **Glass Syndrome or 2q33.1 Microdeletion Syndrome.**
- SAS is a **rare genetic neurodevelopmental disorder** caused by alterations of the SATB2 gene on the second chromosome.
- The **SATB2 gene was discovered in 2003** and **first cases** were reported around **2015**.
- This syndrome significantly affects many organ systems and thus areas of development, primarily brain, palate, teeth and bones.
- A recent study estimated SAS to be **among top 10 monogenic** neurodevelopmental disorders.
- In our current SAS community, the **majority are children and young adults, but the range is from prenatal to 50 years.**

THERE ARE AROUND 800 SAS INDIVIDUALS IDENTIFIED IN 50 COUNTRIES WORLDWIDE. ABOUT HALF OF THEM ARE IN EUROPE.

TO DATE THERE ARE NO SYNDROME-SPECIFIC TREATMENTS OR CURES.

**DESPITE THEIR MANY CHALLENGES,
SATB2 KIDS ARE BUNDLES OF JOY AND DETERMINATION!
WE ARE WORKING RELENTLESSLY TO ENABLE THEM A LIFE WITHOUT SUFFERING AND A CHANCE TO BE AS INDEPENDENT AS POSSIBLE.**

TO LEARN MORE VISIT SATB2 PORTAL



WHAT ARE THE MAIN STRUGGLES OF SAS INDIVIDUALS?

Children and adults with SAS face many challenges that make if **difficult for them to socialise** and, above all, to **live independently in adulthood**. The main obstacles are absent or limited speech, reduced intellectual and motoric ability and autistic spectrum behaviors or other challenging behaviours.

ARE THERE ANY HELPFUL TREATMENTS, INTERVENTIONS?

There are **no syndrome-specific treatments**, but there are clinical interventions recommended to treat the symptoms of SAS, including **intensive speech** (with the focus on **enabling communication** via alternative communication, sign language, gestures)**, occupational and physical therapies.** Commonly used medications today are aimed at **ameliorating epilepsy, sleep disturbances, challenging behaviour, low bone density and constipation**. There are no official clinical guidelines for SAS yet, but **we are working hard with the global SAS community to provide one.**

IS THERE A REFERENCE CENTER FOR SAS IN EUROPE?

There are no centres of expertise for SAS in Europe just yet, but **we are very close to establish one at Radboud Nijmegen, the Netherlands**. There are 2 multidisciplinary clinics though - one in Arkansas, USA and one in London, UK (only for English patients).

ARE THERE ANY SAS PATIENT ORGANISATIONS?

Yes there are 3 international patient organisations:
SATB2 Europe, **SATB2 Gene Foundation**, **USA** and **SATB2 Connect**, **Australia & NZ**.
There are 5 national organisations in Europe - in **Spain**, **France**, **UK**, **Netherlands & Belgium** and **Italy**.
Plus other not yet formal SAS organisations in Poland and Brasil.
Families are welcome to contact their closest organisations and to join international Facebook group!

HOW CAN YOU GET INVOLVED?

- **Parents of SAS individuals** - share your rare journey through different channels and raise awareness in your local, national and global community.
- **Clinicians and researchers** - if you are keen to research and unravel the SATB2 syndrome pathology and contribute your piece in the discovery of possible interventions, we are here to help with your research! Our children need you!
- **You know someone with SAS or simply want to help** - you are welcome to contribute to our cause by a **donation** of any value. **THANK YOU!**