Sturge Weber Syndrome

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What is Sturge-Weber Syndrome?

Sturge-Weber Syndrome (SWS), also known as **encephalotrigeminal angiomatosis**, is a rare congenital disorder characterised by **neurological and dermatological abnormalities**. It is typically identified by a **facial birthmark** called a **port-wine stain** and **vascular malformations** affecting the **leptomeninges** of the brain. This syndrome results from a **somatic mutation in the GNAQ gene**, leading to **abnormal development of capillaries and blood vessels**. SWS is **sporadic**, which means it is not inherited, and it is usually evident at birth. The condition affects approximately 1 in 50,000 live births, presenting with a range of symptoms such as seizures, glaucoma, and potential intellectual disabilities.

What parts of the brain are affected?

In individuals with Sturge-Weber Syndrome, the brain's leptomeningeal angiomatosis primarily impacts the **cerebral cortex**, **particularly on the same side as the facial port-wine stain**. This vascular malformation causes **cortical atrophy and calcifications**, often seen as **“tram-track” patterns in CT scans**. The affected regions correspond to the side of the body where neurological symptoms, like seizures or hemiparesis, manifest. These vascular abnormalities in the brain can lead to decreased cerebral blood flow, contributing to neurological symptoms and complications.

What is the neuropsychological profile of somebody with the syndrome?

The neuropsychological profile of people with Sturge-Weber Syndrome varies widely, depending on the severity and localisation of the brain involvement. Common cognitive and behavioural issues include **developmental delays, learning disabilities, and challenges with attention and executive functioning**. While some individuals may experience intellectual disabilities, this is not universal. Effective management of seizures is crucial, as frequent or uncontrolled seizures

can worsen cognitive impairments. Emotional and behavioural challenges, such as **difficulties in social interaction and adaptive behaviours**, might also occur, affecting educational and social outcomes.

Can patients recover from Sturge-Weber syndrome?

Sturge-Weber Syndrome is a **lifelong condition** with no known cure. However, managing symptoms can significantly enhance quality of life. The prognosis varies among individuals, depending on the severity of neurological involvement, the effectiveness of seizure control, and the presence of other complications such as glaucoma. Early intervention and tailored therapeutic strategies can mitigate some developmental and cognitive impacts, although complete recovery from the syndrome itself is not possible.

Which rehabilitation or compensatory strategies would help?

Rehabilitation and management strategies for Sturge-Weber Syndrome focus on symptom management and quality of life enhancement. Key strategies include:

1. **Seizure Management**: Anticonvulsant medications are primary in controlling seizures, with surgical interventions like hemispherectomy considered when medication is ineffective.
2. **Glaucoma Treatment**: Regular monitoring and treatments, including medications or surgery, are essential to manage increased intraocular pressure and prevent vision loss.
3. **Educational Support**: Individualised educational plans addressing learning difficulties and promoting cognitive development are crucial, often requiring special education services.
4. **Behavioural Interventions**: Therapies such as occupational or speech therapy can help improve functional abilities and communication skills, with behavioural therapies addressing social and adaptive challenges.
5. **Physical Therapy**: For those with motor impairments or hemiparesis, physical therapy aids in improving muscle strength, coordination, and overall mobility.

Early diagnosis and a multidisciplinary approach to care, involving neurologists, ophthalmologists, and educational specialists, are vital for optimising outcomes and supporting individuals with Sturge-Weber Syndrome throughout their lives.

References

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