



For individuals diagnosed with
Smith-Magenis syndrome

Health and Physical Wellbeing

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People with SMS may have a number of physical health needs that benefit from early identification, regular monitoring, and multidisciplinary support. Understanding these common health issues can help families and professionals anticipate and manage problems more effectively.

- **Ears, Nose & Throat:** Recurrent ear infections and hearing loss are common. Hearing should be tested regularly.
- **Vision:** Eye conditions such as myopia, cerebral visual impairment (CVI) or strabismus (squint) are not uncommon. Annual vision checks are recommended.
- **Gastrointestinal problems:** Feeding issues, reflux, and constipation often require ongoing management.
- **Sleep:** Many children and adults with SMS experience disrupted sleep-wake cycles, early waking, and daytime sleepiness.
- **Teeth and Mouth:** Dental crowding, enamel issues, and oral sensitivity may affect eating and hygiene routines.
- **Growth and Development:** Some children may have short stature, delayed puberty, or growth concerns that benefit from regular monitoring.
- **Mobility and Muscles:** Hypotonia, joint laxity, and orthopaedic issues may affect balance, posture, and coordination.

Other Health Concerns

In addition to the more commonly seen health issues, some individuals with SMS may have additional medical conditions. These are more variable and may not affect every person, but they are worth being aware of:

- Scoliosis in individuals with SMS is estimated to be around 30% to 65%
- Cardiac defects – present in approximately 37% of individuals
- Renal abnormalities – around 35% have issues affecting the kidneys, bladder, ureters, or urethra
- Thyroid abnormalities – seen in roughly 29%, including underactive or overactive thyroid
- Low antibody levels – observed in around 23%, which may increase susceptibility to infections
- Seizures – occur in approximately 11–30% of people with SMS
- Abnormal EEG without seizures – found in about 21%
- Facial clefts – including cleft lip or palate, found in around 9%

Not all of these conditions will be present, but early detection and regular reviews can help manage risks effectively. Families should raise any relevant concerns with their GP, paediatrician, or healthcare provider and seek referral to appropriate specialists if symptoms arise.

What Might Help

- Ask your GP for an annual health check, especially if your child or adult is on the learning disability register.
- Keep records of health appointments, reports, and recommendations. A health passport can be a helpful tool.
- Be proactive about vision and hearing checks—many problems are treatable when caught early.
- Talk to your GP about referrals to community paediatrics, physiotherapy, occupational therapy, and dietetics if needed.
- If sleep is a significant issue, request advice from a paediatrician or referral to a sleep service such as children's neurosciences department at Evelina hospital. Medication such as melatonin may be considered with appropriate clinical oversight.

Note for Professionals

A confirmed diagnosis should be followed up with referrals to community paediatrics and allied health professionals (e.g. SALT, OT).

Healthcare needs can evolve over time. Coordinate with families to ensure regular reviews, timely referrals, and a clear communication plan between services.

Annual Health Check (via GP)

Individuals with SMS are eligible for the NHS Annual Health Check (for people with a learning disability) from age 14. This check should include:

- ✓ A review of medical history and medications
- ✓ Physical examination (weight, height, BMI, blood pressure, pulse)
- ✓ Blood tests (e.g. thyroid function, cholesterol, liver and kidney function, blood sugar)
- ✓ Mental health and behaviour screening
- ✓ Vaccination status check
- ✓ Review of vision, hearing, dental and foot care needs
- ✓ Health promotion advice (diet, sleep, physical activity)
- ✓ Carer and family well-being check
- ✓ Update of the health action plan

Birt-Hogg-Dubé syndrome (BHD)

BHD is a rare condition linked to the FLCN gene, which may be affected in some people with Smith-Magenis syndrome. This can increase the risk of kidney tumours, lung cysts or collapsed lung, and skin lesions in adulthood. Screening is usually advised for those with the SMS chromosomal deletion, starting in late adolescence or early adulthood, under the guidance of clinical genetics.

About Us

The SMS Foundation UK is a small national charity supporting families and individuals living with Smith-Magenis syndrome (SMS).

We provide trusted information, practical guidance, and emotional support to help families navigate the challenges of life with SMS.

Our work includes family events, awareness training, education support, and advocating for better recognition and services. Run by a small team with lived experience and a strong network of volunteers, we're here to walk alongside families at every stage of their journey.

Contact Information



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