

Health Problems in Smith-Magenis Syndrome

A range of health difficulties are reported in Smith-Magenis syndrome

Vision (including myopia, both with and without retinal detachment) and hearing problems are frequently noted and infantile hypotonia (lack of muscle tone) and scoliosis (curvature of the spine) are also common.

Peripheral neuropathy is when damage to the nerves at the peripheries (i.e. fingers and toes) causes unusual sensations or numbness in these areas. It has been found in over 55-75% of individuals with Smith-Magenis syndrome and has been associated with self-injurious behaviour.

Other health problems are more variable in nature, including:

- cardiac defects (37%),
- renal abnormalities abnormalities associated with the kidneys, bladder, ureters and urethra (35%),
- thyroid abnormalities (29%),
- low antibodies (23%),
- seizures (11-30%),
- abnormal EEG without seizures (21%),
- forearm abnormalities (16%),
- facial clefts (9%).

For information on how to identify pain in children with severe intellectual disabilities and/or communication difficulties please <u>click here.</u>

Further information on health in Smith-Magenis syndrome can be found on these websites:

Smith-Magenis Foundation

Parents and Researchers Interested in Smith-Magenis Syndrome

National Center for Biotechnology Information