

Health Watch Table — Smith-Magenis Syndrome

Forster-Gibson and Berg 2011

CONSIDERATIONS	RECOMMENDATIONS
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
<p>Children and Adults: Vision: ~ 85% have eye abnormalities, including strabismus, myopia, iris anomalies, and microcornea</p> <p>Retinal detachment, which may be related to self-injurious behaviour in childhood, can occur ~ 25% of adults develop retinal detachment</p> <p>Hearing: Chronic ear infections and hearing loss are common</p> <p>Throat: Almost all have delayed speech ~ 65% have palatal abnormalities such as velopharyngeal insufficiency (VPI) and cleft palate</p> <p>A deep, hoarse voice is common</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Refer to an ophthalmologist following initial diagnosis and annually thereafter <input type="checkbox"/> Arrange an annual hearing assessment during childhood then as per DD Guideline 11 ¹ <input type="checkbox"/> Refer to an ENT surgeon regarding palatal abnormalities following initial diagnosis and annually thereafter <input type="checkbox"/> Refer to a speech and language pathologist in early childhood <input type="checkbox"/> Consider referring to an occupational therapist (OT) or physiotherapist (PT) regarding oral sensorimotor development
2. DENTAL	
<p>Children and Adults: ~ 75% have dental anomalies including tooth agenesis, premolars and taurodontism</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Arrange early and regular dental assessments <input type="checkbox"/> Review brushing and flossing techniques with each dental cleaning
3. CARDIOVASCULAR	
<p>Children & Adults: ~ 50% have congenital cardiovascular abnormalities</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Obtain an echocardiogram <input type="checkbox"/> Refer to a cardiologist at initial diagnosis with follow up arrangements with congenital heart disease clinics, depending on the abnormalities detected. <input type="checkbox"/> Follow recommendations for adults as per DD Guideline 13 ¹
4. RESPIRATORY	
<p>Children & Adults: ~ 75% have sleep disturbances usually related to inverted circadian rhythm of melatonin release</p> <p>Melatonin and acebutolol have been used with some success. Over-the-counter melatonin dosages may be inexact and acebutolol use has some contraindications ²</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake a sleep assessment with attention to sleep disturbance, short sleep cycle, early rising, frequent night awakenings, and daytime napping <input type="checkbox"/> Consider evening melatonin and morning acebutolol (presumed to counter daytime melatonin release) <input type="checkbox"/> Consider strategies to address nighttime safety issues (e.g. enclosed bed) <input type="checkbox"/> If there is evidence of obstructive sleep apnea (OSA), arrange a sleep study
5. GASTROINTESTINAL	
<p>Children and Adults: Feeding problems and gastro-esophageal reflux disease (GERD) are common</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Undertake a clinical assessment in infancy with attention to feeding problems and evidence of GERD <input type="checkbox"/> Monitor regularly for constipation and manage proactively
6. GENITOURINARY	
<p>Children and Adults: Congenital renal or urinary tract abnormalities are common</p> <p>Nocturnal enuresis is common in children</p>	<ul style="list-style-type: none"> <input type="checkbox"/> Obtain a renal ultrasound at initial diagnosis <input type="checkbox"/> Screen for urinary tract infections with an annual urinalysis or as indicated

CONSIDERATIONS	RECOMMENDATIONS
7. MUSCULOSKELETAL	
Children & Adults: ~ 75% of children develop scoliosis, which tends to become more severe with age	<input type="checkbox"/> Obtain spine X-rays at diagnosis to assess for vertebral anomalies then annually to assess for scoliosis
8. NEUROLOGY	
<p>Children: ~ 90% have speech and motor delay as well as hypotonia (particularly in infancy) ~ 75% have peripheral neuropathy, often associated with decreased pain sensitivity</p> <p>Hereditary neuropathy with liability to pressure-related palsies may occur in those with relatively large chromosomal deletions</p> <p>~ 10% -30% have evident and subclinical epilepsy</p>	<input type="checkbox"/> Undertake a neurological assessment at diagnosis and annually thereafter as clinically indicated <input type="checkbox"/> Provide periodic neurodevelopmental assessments during infancy and childhood <input type="checkbox"/> Arrange speech and language pathologist, PT and OT assessments in infancy and periodically thereafter as appropriate <input type="checkbox"/> Consider subclinical seizures if behaviour change occurs <input type="checkbox"/> To evaluate seizures, consider electroencephalography (EEG), and Computed Axial Tomography (CAT) scan and Magnetic Resonance Imaging (MRI) scan of head as indicated during infancy and childhood
9. BEHAVIOURAL/MENTAL HEALTH	
<p>Children & Adults: Self-injurious, maladaptive, and other behaviours (e.g., head banging, nail yanking, self-hugging, teeth grinding, and inserting objects into body orifices) are nearly always present</p> <p>These may decrease with time</p>	<input type="checkbox"/> In children, arrange early intervention with specific preventative behavioural strategies and special education techniques that emphasize individualized instruction <input type="checkbox"/> Use of computer-assisted technology and medication may be helpful <input type="checkbox"/> An annual interdisciplinary team assessment of children is warranted and may also be helpful for adults <input type="checkbox"/> Plan respite care, family psychological and social supports <input type="checkbox"/> Facilitate contact with Parents and Researchers interested in Smith-Magenis Syndrome (PRISMS) to provide support and education (see website below)
10. ENDOCRINE	
<p>Children and Adults: ~ 25% are mildly hypothyroid</p> <p>Hypercholesterolemia is common</p> <p>Hypoadrenalism, though rare, can occur, particularly in children</p>	<input type="checkbox"/> Undertake annual thyroid function and fasting lipid testing <input type="checkbox"/> Start screening for hypercholesterolemia in childhood and consider dietary modification for hypercholesterolemia and the possible role of medication <input type="checkbox"/> Assess for hypoadrenalism in the event of any serious illness
11. INFECTIOUS DISEASE / IMMUNIZATION	
Children & Adults: IgA is reduced in some	<input type="checkbox"/> Arrange qualitative immunoglobulin testing at diagnosis <input type="checkbox"/> Undertake periodic review if recurrent infections
12. OTHER	
<p>Children and Adults: Phenotype/genotype correlations are beginning to emerge for 17p11.2 deletions of different size and for RAI1 mutation carriers</p> <p>Relatively rare condition, first described in the 1980s, may be under recognized</p> <p>Limited data and recommendations are currently available for adults but more information is emerging as identified children age</p>	

Resources

Six published Smith-Magenis syndrome health care guidelines reviewed and compared. (For full list of references see: www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx.)

Smith Magenis syndrome website that may be helpful for families and caregivers

www.prisms.org is a website for Parents and Researchers interested in Smith-Magenis syndrome or google “PRISMS”.

Developed by: *Forster-Gibson, Cynthia, MD, PhD; Berg, Joseph M, MB, BCh, MSc, FRCPSYCH, FCCMG*

Expert Clinician Reviewer

Thanks to the following clinician for her review and helpful suggestions:

Kerry Boyd, MD
McMaster Children’s Hospital,
Hamilton Health Sciences, Hamilton, Ontario
Chief Clinical Officer, Bethesda Services, Thorold, Ontario

References

1. Sullivan WF, Berg JM, Bradley E, Cheetham T, Denton R, Heng J, Hennen B, Joyce D, Kelly M, Korossy M, Lunskey Y, McMillan S. Primary care of adults with developmental disabilities: Canadian consensus guidelines. *Can Fam Physician* 2011;57:541-53.
2. De Leersnyder H, de Blois MC, Bresson JL, Sidi D, Claustrat B, Munnich A. Inversion of the circadian melatonin rhythm in Smith-Magenis syndrome. *Rev Neurol (Paris)*. 2003 Nov;159(11 Suppl):6S21-6.