

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

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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

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Kerry Boyd, MD
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References

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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.