Health Watch Table — Smith-Magenis Syndrome

Forster-Gibson and Berg 2011

CONSIDERATIONS	RECOMMENDATIONS
1. HEENT (HEAD, EYES, EARS, NOSE, THROAT)	
Children and Adults: Vision: ~ 85% have eye abnormalities, including strabismus, myopia, iris anomalies, and microcornea Retinal detachment, which may be related to self-injurious behaviour in childhood, can occur ~ 25% of adults develop retinal detachment Hearing: Chronic ear infections and hearing loss are common Throat: Almost all have delayed speech ~ 65% have palatal abnormalities such as velopharyngeal insufficiency (VPI) and cleft palate A deep, hoarse voice is common	☐ Refer to an ophthalmologist following initial diagnosis and annually thereafter ☐ Arrange an annual hearing assessment during childhood then as per DD Guideline 11 [1] ☐ Refer to an ENT surgeon regarding palatal abnormalities following initial diagnosis and annually thereafter ☐ Refer to a speech and language pathologist in early childhood ☐ Consider referring to an occupational therapist (OT) or physiotherapist (PT) regarding oral sensorimotor development
2. DENTAL	
Children and Adults: ~ 75% have dental anomalies including tooth agenesis, premolars and taurodontism	☐ Arrange early and regular dental assessments ☐ Review brushing and flossing techniques with each dental cleaning
3. CARDIOVASCULAR	
Children & Adults: ~ 50% have congenital cardiovascular abnormalities	☐ Obtain an echocardiogram ☐ Refer to a cardiologist at initial diagnosis with follow up arrangements with congenital heart disease clinics, depending on the abnormalities detected. ☐ Follow recommendations for adults as per DD Guideline 13 [1]
4. RESPIRATORY	
Children & Adults: ~ 75% have sleep disturbances usually related to inverted circadian rhythm of melatonin release Melatonin and acebutolol have been used with some success. Over-the-counter melatonin dosages may be inexact and acebutolol use has some contraindications [2]	☐ Undertake a sleep assessment with attention to sleep disturbance, short sleep cycle, early rising, frequent night awakenings, and daytime napping ☐ Consider evening melatonin and morning acebutolol (presumed to counter daytime melatonin release) ☐ Consider strategies to address nighttime safety issues (e.g. enclosed bed) ☐ If there is evidence of obstructive sleep apnea (OSA), arrange a sleep study
5. GASTROINTESTINAL	
Children and Adults: Feeding problems and gastro-esophageal reflux disease (GERD) are common	☐ Undertake a clinical assessment in infancy with attention to feeding problems and evidence of GERD ☐ Monitor regularly for constipation and manage proactively
6. GENITOURINARY	
Children and Adults: Congenital renal or urinary tract abnormalities are common Nocturnal enuresis is common in children	☐ Obtain a renal ultrasound at initial diagnosis ☐ 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	☐ Obtain spine X-rays at diagnosis to assess for vertebral anomalies then annually to assess for scoliosis
8. NEUROLOGY	
Children: ~ 90% have speech and motor delay as well as hypotonia (particularly in infancy) ~ 75% have peripheral neuropathy, often associated with decreased pain sensitivity Hereditary neuropathy with liability to pressure-related palsies may occur in those with relatively large choromosomal deletions ~ 10%-30% have evident and subclinical epilepsy	☐ Undertake a neurological assessment at diagnosis and annually thereafter as clinically indicated ☐ Provide periodic neurodevelopmental assessments during infancy and childhood ☐ Arrange speech and language pathologist, PT and OT assessments in infancy and periodically thereafter as appropriate ☐ Consider subclinical seizures if behaviour change occurs ☐ 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	
Children & Adults: Self-injurious, maladaptive, and other behaviours (e.g., head banging, nail yanking, self-hugging, teeth grinding, and inserting objects into body orfices) are nearly always present These may decrease with time	☐ In children, arrange early intervention with specific preventative behavioural strategies and special education techniques that emphasize individualized instruction ☐ Use of computer-assisted technology and medication may be helpful ☐ An annual interdisciplinary team assessment of children is warranted and may also be helpful for adults ☐ Plan respite care, family psychological and social supports ☐ Facilitate contact with Parents and Researchers interested in Smith-Magenis Syndrome (PRISMS) to provide support and education (see website below)
10. ENDOCRINE	
Children and Adults: ~ 25% are mildly hypothyroid Hypercholesterolemia is common Hypoadrenalism, though rare, can occur, particularly in children	☐ Undertake annual thyroid function and fasting lipid testing ☐ Start screening for hypercholesterolemia in childhood and consider dietary modification for hypercholesterolemia and the possible role of medication ☐ Assess for hypoadrenalism in the event of any serious illness
11. INFECTIOUS DISEASE / IMMUNIZATION	
Children & Adults: IgA is reduced in some	☐ Arrange qualitative immunoglobulin testing at diagnosis ☐ Undertake periodic review if recurrent infections
12. OTHER	
Children and Adults: Phenotype/genotype correlations are beginning to emerge for 17p11.2 deletions of different size and for RAI1 mutation carriers Relatively rare condition, first described in the 1980s, may be under recognized Limited data and recommendations are currently available for adults but more information is emerging as identified children age	

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".

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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.

