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	☐ Obtain a renal ultrasound at initial diagnosis.

CONSIDERATIONS	RECOMMENDATIONS
urinary tract abnormalities are common Nocturnal enuresis is common in children	☐ Screen for urinary tract infections with an annual urinalysis or as indicated.
7. MUSCULOSKELETAL (MSK)	
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 chromosomal 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. BEHAVIOIURAL/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 orifices) 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.	

WEBSITES 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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