# Health Watch Table – Smith-Magenis Syndrome

*Forster-Gibson and Berg 2011*

<table>
<thead>
<tr>
<th>CONSIDERATIONS</th>
<th>RECOMMENDATIONS</th>
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<tr>
<td>1. <strong>HEENT (HEAD, EYES, EARS, NOSE, THROAT)</strong></td>
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</table>
| Children and Adults: Vision: ~ 85% have eye abnormalities, including strabismus, myopia, iris anomalies, and microcornea | □ Refer to an ophthalmologist following initial diagnosis and annually thereafter.  
 □ Arrange an annual hearing assessment during childhood then as per DD Guideline 11.  
 □ 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. |
| 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 |  |
| 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. |
| 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 | □ 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. |
<p>| 6. <strong>GENITOURINARY</strong> |  |
| Children and Adults: Congenital renal or | □ Obtain a renal ultrasound at initial diagnosis. |</p>
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<td>urinary tract abnormalities are common</td>
<td>□ Screen for urinary tract infections with an annual urinalysis or as indicated.</td>
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<tr>
<td>Nocturnal enuresis is common in children</td>
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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. **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 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”.

REFERENCES CITED


PUBLISHED HEALTH CARE GUIDELINES REVIEWED AND COMPARED


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About this Health Watch Table

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