GUIDELINES FOR THE MANAGEMENT OF CHILDREN AND ADULTS WITH SMITH-MAGENIS SYNDROME

These guidelines were developed by the Scientific and Clinical Advisory Group of the Smith-Magenis Syndrome Foundation, U.K. They are aimed at health care professionals working with individuals with Smith-Magenis Syndrome, and will be reviewed and updated at regular intervals.

Over and above the routine health and vaccination schedules administered to all children, we recommend consideration of the following:

ON DIAGNOSIS:

1. PHYSICAL AND NEUROLOGICAL ASSESSMENT AND EXAMINATION

CLINICAL AREA	INVESTIGATION/ASSESSMENT	REASON
GROWTH, FEEDING & NUTRITION	Height and weight centiles Feeding evaluation – examination of palate, assessment of swallowing, oral motor skills, gastroesophageal reflux, caloric intake. Referral to specialist feeding team if indicated	short stature and/or failure to thrive in 78% of cases; tracheobronchial problems in 50%).
CARDIAC	cardiac examination including echocardiogram,	Cardiac problems in over 30%.
RENAL	Renal ultrasound	renal/urologic anomalies in over 30% of cases
IMMUNE SYSTEM	Assessment of Immune function if presenting with frequent infections	decreased IgG or IgA in over 30% of cases).
HEARING	Assessment for conductive and or sensorineural hearing loss. Grommets or hearing aid may be indicated.	60% have hearing problems – 65% conductive, 35% sensorineural
EARS, NOSE, THROAT	Otolaryngologic evaluation to assess ear, nose and throat problems, with specific attention to ear physiology and palatal abnormalities	velopharyngeal insufficiency in 75%, cleft in 10%).
EYES	Ophthalmologic examination with attention to evidence of strabismus, microcornea, refractive error, retinal detachment.	
SPINE	Assessment for scoliosis	in over 60% of those aged 4 years and over

GENETICS	Individuals with larger deletions → screen for adrenal function → assessment of nerve conduction velocity if the gene PMP22 is involved.	gene PMP22 is involvement associated with hereditary neuropathy with liability to pressure palsy.

2. GENETICS

Referral to Clinical Geneticist for parental chromosome analysis and provision of genetic counselling.

3. SLEEP DIFFICULTIES

Sleep history to document the sleep cycle and evidence for sleep apnea, snoring and other signs of sleep abnormalities. Actigraphy, if available, is a useful non-invasive method of capturing sleep-wake cycles in the home environment over a period of time. Any concerns about snoring or irregular night-time breathing should prompt a full polysomnography and ENT evaluation.

4. BEHAVIOUR DIFFICULTIES

Consider referral to Clinical Psychology or Learning Disability CAMHS services for detailed assessment and intervention.

5. FAMILY SUPPORT

The child's/adult's behaviour and sleep difficulties may be chronic, and in many cases support from Social Services Learning Disability Services, Child Development Team staff (e.g. keyworker), Child and Adolescent Mental Health Services, Voluntary Organisations and Parent Support Groups may be helpful.

ANNUAL ASSESSMENT

- Monitor for scoliosis
- Thyroid Function.
- Consider ophthalmologic evaluation.
- Audiologic evaluation at regular intervals or as clinically indicated to monitor for conductive or sensorineural hearing loss.
- Periodic assessment of presence of challenging behaviours and referral for treatment as needed.

Management of Sleep Difficulties
Management of sleep disorders is likely to include behaviour management, and may also include melatonin at night and possibly beta blockers or phototherapy in the morning. The timing of such interventions is likely to be very important and may vary from child to child. No formal controlled trials of these latter interventions have been conducted to date. However, they are the subject of much research interest and discussion. Referral to a specialist sleep service might be indicated.

Management of Challenging Behaviours
For challenging behaviours, including aggression, self injurious behaviours and impulsivity/hyperactivity, both conventional behaviour therapy and pharmacological treatments should be considered, though these behaviours are often very difficult to treat.

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