Smith Magenis Syndrome: Challenge to Services

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Smith – Magenis Syndrome

Drs. Anne Smith & Elen Magenis (1986)

Prevalence: 1:25,000 M = F

Microdeletion: short arm Ch17

Gene: Retinoic acid-induced protein 1 (RAI1)

Occurrence is de novo

Behavioural Phenotype

Cognitive ability: mild to moderate

Life expectancy: approximates to general population

SMS: Physical Features

Facial appearance: flattened mid-face, down-turned mouth, “Cupid’s lips” prominent cheeks and jaw; eyebrows meet in midline (synophrys)

Middle ear problems (chronic ear infections) and laryngeal anomalies

Hoarse voice, hypernasal speech

Short fingers and toes; flat feet; broad-based gait

Hyporeflexia

Decreased sensitivity to pain

Development & Behaviour

(> 75% of affected individuals)

Developmental delay
Generalized complacency / lethargy (infancy)
Mouthing objects beyond early childhood
Speech delay and articulation problems
Sensory integration difficulties
Teeth grinding
Delayed toileting skills

Behaviours: hyperactivity, impulsivity, attention seeking (especially from adults); excitability or distractibility;
sudden mood shifts; explosive outbursts; prolonged tantrums; destructive behaviour

Development & Behaviour
(> 75% of affected individuals)

Sleep disturbance (chronic)
- **Frequent night time waking**
- **Increased daytime sleepiness / naps**
- **Early morning waking**

Stereotypic / repetitive behaviours:
- **self-hugging / hand squeezing**

Self-injurious behaviours: head banging, hand biting, picking skin, pulling off finger and toenails

Inserting foreign objects into body orifices

Positive behavioural features
- Endearing personality; memory; sense of humour

Behavioural Phenotype

A characteristic pattern of motor, cognitive, linguistic and social abnormalities which is consistently associated with a biological disorder (SSBP UK 2007)

The concept of behavioural phenotypes is intended to form a basis for research into behavioural, emotional, and other aspects of biologically determined syndromes associated with intellectual disability or mental retardation. The aim is to encourage research and information sharing.

Case Study

31 year old woman

Mild Intellectual Disability

Verbal \ physical aggression

“Control + Restraint”

Strong attachments to males

Mood swings: extreme \ rapid

Sleep Pattern: erratic

Concentration: poor

Speech: sexual content

Impacts

Effect on residents: dictates the atmosphere

Effect on staff: monopolises attention

Effect on community access: self and others

Effect on property

Effect on service: alienation from staff and residents

Development

Delayed developmental milestones

Behaviour: temper tantrums from 4 years; aggression

Schooling: learning difficulties; special needs school

Social care: fostered at 4 years – failed; care home at 6 years

Residential school 10 – 19 years

No employment

No sexual relationships

No known history of abuse

Family History

Second eldest of four siblings

Fostered at the age of four

No documented family history of mental illness

Psychiatric History

Behaviour: assaulting staff, throwing objects, breaking windows, stripping naked, punching, kicking, spitting, slapping faces, incontinent of urine, absconding

Admissions (1999 – 2006): MHA x3; Informal x2

Opinion of MDT: "few facilities that could cope with behaviour and meet her needs"

No forensic history

Drug History

Quetiapine 750mg

Haloperidol 15mg

Benperidol 1.5mg

Haloperidol PRN

Semi-Sodium Valproate 1.5g

Clonazepam 3mg

Lorazepam PRN

Medical History

BMI > 50   Wt. 126kg

Hearing impairment

Hypoplasia of the maxilla

Multiple dental caries

Abnormal gait

Mental State Examination

Obese, caucasian woman, hirsute, grinding teeth

Head shaking and nodding, self-hugging, squeezing hands together, loud and hoarse voice

No signs of mood disorder or psychosis

Behaviours as In-patient

Demanding of staff

Urinary incontinence

Intense attachment

Domineering others

Excessive eating

Sleep pattern

Sexual behaviours – alleges male nurses are her boyfriends

Intense rage: PRN medication

Summary

31 year old female: Smith-Magenis Syndrome; mild ID

Failed community placement

In-patient for 3 years - severe challenging behaviour

No signs of mental illness

Residential schools

Hospital admissions under MHA

No family support

Assessment

Behaviour: sleep; urinary incontinence; oppositional; eating; sexual

Physical assessment: weight; hearing; teeth; sensation

Mental health assessment

Review medication

Intervention

Behaviour

Attachment 1:1 support

Social activities

Drug Therapy

Rationalise drug regimen

- Reduce anti-psychotics;
- Stop benzodiazepines
- Mood stabiliser – Lithium Carbonate

Measure

HoNOS – LD

Incident Reports

Incident Reports

2006	159
2007	110
2008	45
2009	14

HoNOS – LD Scores

Date	Score	Intervention
Jan 2007	33	Reduce Clonazepam
May 2007	22	Reduce HLP
July 2007	12	Reduce VALP
Dec 2007	9	Maintain
2008	15	Lithium Carnbonate
2009	8	Lithium Carbonate Maintenance

Clinical Achievements

Rationalise drug regimen

Haloperidol 5mg

Lithium Carbonate 1g

Withdraw: Clonazepam; Benperidol; Quetiapine; Semi Soduium Valproate

Behaviour: sleep routine; bladder routine; attachments

Day - time activities: music therapy; beauty sessions

Social: holiday; community outings

Lessons Learned

Diagnosis: Phenotype \ Illness

Use of Medication

Behavioural Management

Meaningful Life Activities

Placement

Resources

Udwin O (2003)	Cambridge UP
Gene Reviews	www.geneclinics.org
SSBP	www.ssbp.co.uk
SMS Foundation	www.smith-magenis.co.uk