Hyperacusis in Smith-Magenis Syndrome

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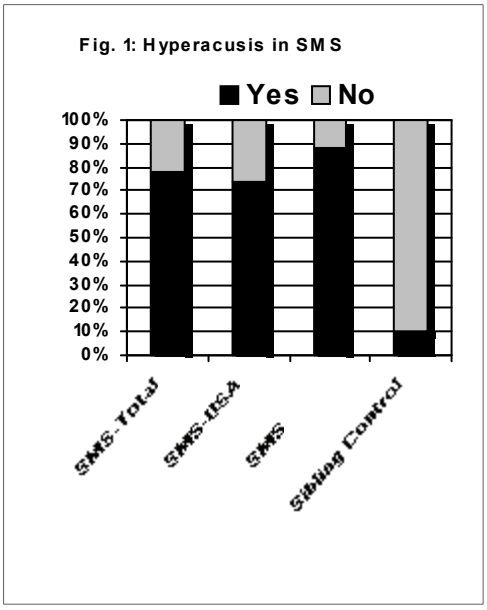
This research was conducted by audiology doctoral student, Ms. Jennifer Bentley working with audiologists Carmen Brewer, PhD and Chris Zalewski, MA as part of the IRB-approved comprehensive natural history study of Smith-Magenis syndrome (protocol 01-HG -0109) being conducted at the National Institutes of Health (NIH). This article is based on Dr. Brewer's presentation at the May 2007 PRISMS Conference in Reston, VA.

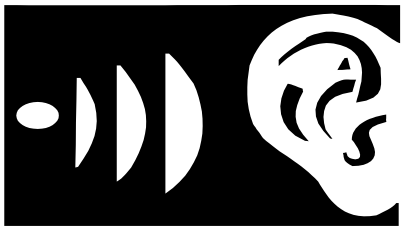
ENT abnormalities are documented in as many as 94% of individuals with Smith-Magenis syndrome (SMS). Most children with SMS experience chronic otitis media that begins in early infancy and hearing loss (conductive, sensorineural, or mixed) is common. Additionally, oversensitivity to loud sounds is an expressed concern of a number of parents.

Hyperacusis is defined as an oversensitivity to certain frequency ranges or certain sounds that are not bothersome to listeners with normal hearing. Due to the similarities between SMS and other genetic syndromes with known hyperacusis, such as William syndrome, NIH researchers sought to investigate this question further.

The purpose of this study was to determine the occurrence and severity of hyperacusis in persons with SMS and to document the types of responses, triggers and management techniques. Children with a confirmed diagnosis of SMS participating in the IRB-approved natural history study of SMS at NIH (01-HG-0109) were recruited in the United States or at Camp Breakaway in Australia. A 2-page questionnaire originally designed to evaluate the severity of hyperacusis in children with Williams syndrome (Cohen et al, 2006) was mailed to 90 families in the USA (SMS-USA) and distributed to 16 families attending Camp-Breakaway (SMS-AUS). Unaffected SMS siblings of the USA group served as a comparison (control) group.

Results: Preliminary findings are summarized below based on analysis of 83 returned questionnaires including: 47 (29M/18F) from SMS-USA, 16 (8M/8F) from SMS-AUS and 20 (13M/7F) from the sibling control group: Sensitivity to loud sound (hypercusis) was present in 78% of the total SMS group compared to only 10% of the sibling control group (Fig 1). While there was a slight difference in the frequency between the SMS-USA group (74%) and SMS-AUS group (87%), this difference was not significant. Thus, in this study, hyperacusis was about 7 times higher in the SMS group. While the majority (59%) of parents indicated that their child's intolerance for loud





sounds remained unchanged over time, 33% felt that it had improved and 2% felt that it had cleared up.

Triggers and Response to Loud Sounds

The major trigger for distress was tiredness (47%) followed by mood (29%). Tiredness was also the most often cited reason for a heightened reaction in 50%. Common behavioral responses to distressing sounds were covering the ears with hands (88%), becoming upset (58%), or displaying anxiety/tension (52%). Self-injurious behaviors (head banging, biting self), common in SMS, were triggered by loud sounds in 28% of the SMS group.

Parents reported the most severe reations for sudden environmental sounds such as fireworks, balloon burst, and a sudden shout. There did not appear to be a relationship between the presence/absence of hyperacusis and the child's degree or type of hearing loss.

Palliative Strategies Tried (See Table 1)

Many parents reported that their child with SMS showed **less** distress when they were prepared or warned of an impending sound. Other effective management techniques included sound reduction through a variety of techniques, avoidance of distressing sounds, controlling known triggers, and familiarization with the offending sound.

Summary: A significant percentage (78%) of children with SMS were reported to have hyperacusis in this initial study. Research is ongoing to characterize this newly appreciated finding in SMS and identify strategies to aid in management. Assessment of hypersensitivity to loud sounds should be included in the audiologic evaluation of persons with SMS. This can be achieved via traditional loudness discomfort measures or by questionnaire. When a problem is identified, parents/caretakers should be counseled regarding palliative strategies.

What Is Smith-Magenis Syndrome?

Smith-Magenis syndrome (SMS) is a chromosomal disorder characterized by a specific pattern of physical, behavioral and developmental features. It is caused by a missing piece of genetic material from chromosome 17, referred to as deletion 17p11.2. The first group of children with SMS was described in the 1980's by Ann CM Smith, MA, a genetic counselor, and Ellen Magenis, MD, a physician and cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in 1 out of 25,000 births. SMS is underdiagnosed, but as awareness of it increases, the number of people identified grows every

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Table 1: Successful Palliative Strategies Tried

Strategy	Number Reporting
Preparation, explanation, warning	14
Avoidance of sounds	4
Control triggers (e.g., nap to avoid fatigue)	2
Reduce sound level	
Earplugs or headphones	5
Cover ears	1
Turn off hearing aids	1
Increase familiarity with sound	2