LETTERS TO THE EDITOR

The SNATIATION reflex

I read with the greatest delight the report by Teebi and Al-Saleh describing possibly one of their own families in keeping with the ACHOO tradition) the autosomal dominant inheritance of a sneezing disorder provoked by fullness of the stomach.

As the authors point out (possibly with tongue in cheek) it may not be all that uncommon a condition, but simply has not been previously reported because it does not lead to major disability. On the other hand it could be socially embarrassing or even stigmatising, albeit highly preferable to a compulsive belching reflex at the end of heavy meals.

The basic epidemiological and genetic questions of frequency, prevalence, selective advantage, presence in other species, pathogenic mechanism, linkage, nucleotide sequence, number of introns (perhaps related to the number of sneezes), transposable elements, etc await further investigation. However, I would like to suggest that a catchy acronym may hasten the process of reporting other families (although it hasn’t helped ACHOO much). Therefore, I propose the newly described condition be called the SNATIATION reflex—a combination of sneezing and satiation and easily remembered by the acronymous handle of Sneezing Non-controllably At a Time of Indulgence of the Appetite—a Trait Inherited and Ordained to be Named.

In all seriousness I really was delighted to see the report, both because it tickled my imagination and because I think it is important to report normal traits both structural and behavioural. We tend to teach human genetics by diseases and have few examples of non-pathological traits determined by single genes. Furthermore, the mechanisms involved in producing the sneezes in both the ACHOO and SNATIATION reflexes are totally unknown. Are there other inherited sneezing reflexes? Time to get busy surveying friends, relatives, and clinic personnel!

JUDITH G HALL
University of British Columbia
Clinical Genetics Unit,
Grace Hospital, Vancouver, Canada.


Angelman’s syndrome

It has taken dysmorphologists a number of years confidently to recognise the facial features of the syndrome characterised by severe mental retardation, marked speech impairment (usually less than three words), jerky movements, and a happy disposition. Angelman’s syndrome is now well established as a clinical entity with a recognisable facial appearance, a distinctive EEG pattern, and a chromosomal deletion of 15q11-13 in 40 to 50% of cases. Indeed it is often possible to suggest the diagnosis from facial photographs.

We wish to draw attention to an error that we have made on two occasions in the past few years which will be of interest to clinicians. Angelman’s syndrome was originally thought to be the diagnosis in the two girls we describe at about 2 years of age. With time the diagnosis of Rett’s syndrome became obvious. Both patients have a similar facial appearance with the subtle facial dysmorphism seen in Angelman’s syndrome, that is, a prominent lower jaw, a wide mouth, and midfacial hypoplasia. The two patients who were erroneously diagnosed as having Angelman’s syndrome at about 2 years of age both tended to tongue thrust, had jerky movements, seizures, and significant global delay with minimal speech. As time went on, regression became apparent with deceleration in the rate of head growth leading to microcephaly; they lost their happy disposition and developed the typical involuntary hand stereotypes of Rett’s syndrome. The similar facial dysmorphism of Angelman’s and Rett’s syndromes may be more readily confused in a girl with Rett’s syndrome who has early developmental delay, contrasting with the classical description of Rett’s syndrome with normal early development.

We advise caution in making the diagnosis of Angelman’s syndrome in girls at about 2 years of age without the commonly associated EEG features and with normal chromosomes. The recurrence risk in Angelman’s syndrome with no chromosomal abnormality may be 25% in contrast with Rett’s syndrome which carries a low recurrence risk.

Figure 1 Case 1 (a) aged 2 years, (b) aged 4 years.
The SNATIATION reflex.

J G Hall

*J Med Genet* 1990 27: 275
doi: 10.1136/jmg.27.4.275

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