

LETTERS

'Nasal' speech—hyper or hypo?

European Journal of Human Genetics (2012) **20**, 367;
doi:10.1038/ejhg.2011.228; published online 7 December 2011

Vergult *et al*¹ present interesting and useful descriptions of three patients with deletions at 12q15. It would be helpful if they could clarify one point, however. The authors refer throughout the paper to 'nasal' speech. It is not clear from the description whether their patients have hypernasal speech or hyponasal speech.

Hypernasal speech occurs when there is escape of airflow and acoustic energy into the nasal cavity during speech, commonly due to dysfunction (mechanical or neuromuscular) of the velopharyngeal valving mechanism, as in velocardiofacial syndrome.² Hyponasal speech, by contrast, is caused by reduced nasal cavity resonance during speech, usually due to anatomical obstruction of the nasal cavity. It is easy to demonstrate, by simply pinching the nose closed while speaking.

Distinguishing between these two abnormalities is important, both in diagnosis and in management of conditions in which they occur.

CONFLICT OF INTEREST

The author declares no conflict of interest.

Edwin P Kirk

Department of Medical Genetics, Sydney Children's Hospital,
Randwick, New South Wales, Australia
E-mail: e.kirk@unsw.edu.au

in our paper,¹ which was published in the October issue of this year, all present with hypernasal speech. For patient 1, the hypernasality was confirmed by the clinician. The hypernasality seen in patient 2 was assessed by the speech and language therapists. No evidence of nasal cavity obstruction was found during examination by an ear, nose and throat surgeon. A velopharyngeal operation was performed on patient 3 due to hypernasality. Such an operation is performed in order to reduce hypernasality, and not for hyponasality. The hypernasal speech is, however, still present in this patient.

Thus, the deletions in our patients seem to be associated with hypernasal speech, hypothyroidism and learning disability or developmental delay.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

Sarah Vergult¹, Danijela Krgovic², Bart Loeys³, Stanislas Lyonnet⁴,
Agne Liedén⁵, Britt-Marie Anderlid⁵, Freddie Sharkey⁶, Shelagh Joss⁷,
Geert Mortier³ and Björn Menten¹

¹Center for Medical Genetics, Ghent University Hospital,
Ghent, Belgium;

²Laboratory of Medical Genetics, University Medical
Centre Maribor, Maribor, Slovenia;

³Department for Medical Genetics, Antwerp, Belgium;
⁴INSERM, Paris, France;

⁵Department of Clinical Genetics and Institution of
Molecular Medicine and Surgery, Centre of Molecular Medicine,
Karolinska Universitetssjukhuset, Stockholm, Sweden;

⁶South East Scotland Cytogenetics Laboratory,
Western General Hospital, Edinburgh, UK;

⁷West of Scotland Regional Genetics Service, Institute of
Medical Genetics, Yorkhill Hospital, Glasgow, UK
E-mail: bjorn.menten@ugent.be

1 Vergult S, Krgovic D, Loeys B *et al*: Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. *Eur J Hum Genet* 2011; **19**: 1032–1037.

2 Dworkin JP, Marunick MT, Krouse JH: Velopharyngeal dysfunction: speech characteristics, variable etiologies, evaluation techniques and differential treatments. *Lang Speech Hear Serv Schools* 2004; **35**: 333–352.

1 Vergult S, Krgovic D, Loeys B *et al*: Nasal speech and hypothyroidism are common hallmarks of 12q15 microdeletions. *Eur J Hum Genet* 2011; **19**: 1032–1037.

Nasal speech in patients with 12q15 microdeletions

European Journal of Human Genetics (2012) **20**, 367;
doi:10.1038/ejhg.2011.230; published online 7 December 2011

As pointed out correctly by Dr Kirk there is a difference between hypernasal speech and hyponasal speech. The three patients described