Psychological Medicine (2012), **42**, 419–426. © Cambridge University Press 2011
doi:10.1017/S0033291711001115

ORIGINAL ARTICLE

Affective functioning and social cognition in Noonan syndrome

E. Wingbermühle[1,2]*, J. I. M. Egger[1,2,3], W. M. A. Verhoeven[1,4], I. van der Burgt[5] and R. P. C. Kessels[1,2,6]

[1] *Vincent van Gogh Institute for Psychiatry, Venray, The Netherlands*
[2] *Donders Institute for Brain, Cognition and Behaviour, Centre for Cognition, Radboud University Nijmegen, The Netherlands*
[3] *Behavioural Science Institute, Radboud University Nijmegen, The Netherlands*
[4] *Department of Psychiatry, Erasmus University Medical Centre, Rotterdam, The Netherlands*
[5] *Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands*
[6] *Department of Medical Psychology, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands*

Background. Noonan syndrome (NS) is a common genetic disorder, characterized by short stature, facial dysmorphia, congenital heart defects and a mildly lowered IQ. Impairments in psychosocial functioning have often been suggested, without, however, systematic investigation in a clinical group. In this study, different aspects of affective processing, social cognition and behaviour, in addition to personal well-being, were assessed in a large group of patients with NS.

Method. Forty adult patients with NS were compared with 40 healthy controls, matched with respect to age, sex, intelligence and education level. Facial emotion recognition was measured with the Emotion Recognition Task (ERT), alexithymia with both the 20-item Toronto Alexithymia Scale (TAS-20) and the Bermond–Vorst Alexithymia Questionnaire (BVAQ), and mentalizing with the Theory of Mind (ToM) test. The Symptom Checklist-90 Revised (SCL-90-R) and the Scale for Interpersonal Behaviour (SIB) were used to record aspects of psychological well-being and social interaction.

Results. Patients showed higher levels of cognitive alexithymia than controls. They also experienced more social distress, but the frequency of engaging in social situations did not differ. Facial emotion recognition was only slightly impaired.

Conclusions. Higher levels of alexithymia and social discomfort are part of the behavioural phenotype of NS. However, patients with NS have relatively intact perception of emotions in others and unimpaired mentalizing. These results provide insight into the underlying mechanisms of social daily life functioning in this patient group.

Received 5 April 2011; Revised 2 June 2011; Accepted 3 June 2011; First published online 11 July 2011

Key words: Alexithymia, behavioural phenotype, emotion perception, genetics, neuropsychology.

Introduction

Noonan syndrome (NS) is an autosomal dominant congenital disorder, first described in the 1960s (Noonan & Ehmke, 1963). Although relatively unknown, NS is fairly common, with an estimated incidence between one in 1000 and one in 2500, and a presumably higher prevalence of milder expression (Allanson, 2007). Different activating germline mutations in genes encoding participants of the Ras-mitogen-activated protein kinase (MAPK) pathway have been found to cause NS. At present, genetic testing reveals a mutation in approximately 70% of Noonan cases. Research aimed at the identification of other NS-causing genes is ongoing and the diagnosis is still established primarily on clinical grounds, by observation of key features such as short stature, heart defects and facial dysmorphology. Typical facial appearance involves ptosis, wide-spaced eyes, low-set and posteriorly rotated ears with a thickened helix, and a broad or webbed neck.

Although cardinal characteristics are well described, there is a large variability in expression and, with increasing age, the phenotype often becomes less pronounced. Diagnosis is generally made in early childhood because of congenital heart disease, feeding problems or other somatic complications.

* Address for correspondence: E. Wingbermühle, M.Sc., Centre of Excellence for Neuropsychiatry, Vincent van Gogh Institute for Psychiatry, Stationsweg 46, 5803 AC Venray, The Netherlands.
(Email: pwingbermuhle@vvgi.nl)