

NOONAN FACT FILE

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BEHAVIORAL ASPECTS AND PSYCHIATRIC FINDINGS IN NS

Abstract

This study describes the behavioural phenotype and psychiatric symptoms of 21 children with Noonan's syndrome attending a paediatrics genetics clinic. Data from the behavioural phenotypes questionnaire are presented that suggest that children with Noonan's syndrome tend to be clumsy, stubborn, irritable, have communication difficulties and are 'faddy eaters'. Nearly 50% of the patients with Noonan's syndrome were psychiatric 'cases' as maintained by a standardised measure.

(Arch Dis Child 1995; 72; 153-155)

Keywords: Noonan's syndrome, behavioural phenotype, psychiatric symptoms.

Noonan's syndrome is a genetic syndrome featuring cardiac abnormalities (chiefly pulmonary stenosis), facial abnormalities, short stature and learning difficulties. Although familial recurrence is consistent with an autosomal dominant mode of inheritance, sporadic cases occur. There is a variability of expression with some transmitting family members having minimal features.¹ The overall incidence is believed to be between one in 1000 and one in 2500.² There are few data on cognition and intelligence in Noonan's syndrome. A study by Money and Kalus of eight males with Noonan's syndrome aged 13-36 years, showed an IQ range from 64-127 with a median of 102.³ Five of these patient showed specific verbal or praxic disparity.

There is an increasing body of knowledge about behavioural phenotypes of different genetic syndromes.⁴ The impetus for this research is derived from parent groups, who often cite their children's behaviour as the least understood and the most difficult to cope with aspect of their genetic condition. It has been observed that children with the same genetic disorder may behave in similar ways. The term 'behavioural phenotype' should be reserved for behaviours which are particularly common in people affected by a give genetic condition and understood as an expression of the genotype. It should not be used as synonymous with a behavioural syndrome or psychiatric diagnosis.⁵ Behavioural phenotypes of different biologically determined mentally handicapping conditions have now been described for Angelman's syndrome,⁶ fragile X syndrome,⁷ tuberous sclerosis, and Williams syndrome.⁹ Most reports of Noonan's syndrome restrict their attention to the psychical characteristics, developmental delay, and attainments. There are few research data available about the behavioural aspects of children with Noonan's syndrome. Hill reports problematic peer relationships, stubbornness, and feeding difficulties in infancy.¹⁰

The identification of behavioural phenotypes has important clinical implications for diagnosis, genetic counselling, and management of children and families with genetic syndromes. The recognition that certain syndromes result in particular behavioural problems helps carers in their understanding and acceptance of their child's condition, facilitates early diagnosis, and alerts the clinician as to the likely need for particular care in designing case management plans. Recent reviews of Noonan's syndrome have commented on areas of limited knowledge including difficulty in making the diagnosis, the lack of a biochemical marker, and uncertainty about recurrence risks.^{1 2 11} Further knowledge of the behavioural and psychiatric characteristics of Noonan's syndrome may improve diagnosis and subsequent management.

This study describes the behavioural phenotype and psychiatric symptoms of 21 children with definite Noonan's syndrome. Data from the behavioural phenotypes questionnaire (BPQ) are presented and, in addition, we asked parents to complete a standard questionnaire of psychiatric morbidity, the child

behaviour checklist (CBCL)¹² for both subjects with Noonan's syndrome and a sibling control group. The study is part of a larger study delineating haematological and genetic characteristics of Noonan's syndrome. As far as we are aware, this is the first published study of both behavioural and psychiatric aspects of the syndrome.

Methods

A total of 27 patients with Noonan's syndrome between the ages of 2 and 19 years were identified from the diagnostic register, department of clinical genetics, Royal Manchester Children's Hospital (RMCH). This is a regional paediatric genetics centre and all cases had been referred for diagnostic assessment and subsequent follow-up. All patients had been assessed by a consultant paediatric geneticist (MS) before the start of this study and all the children easily satisfied the criteria of Allanson *et al* for a diagnosis of Noonan's syndrome.¹ The children were invited to attend with their families for a half day's assessment at RMCH. The first part of this assessment included a detailed clinical examination. Examining the eight syndrome scales of the CBCL, social problems (six cases (32%)) and attention problems (five cases (26%)) were noted for those with Noonan's syndrome. Differences between Noonan's syndrome subjects subscale scores and sibling control scores did not attain statistical significance.

Discussion

This is the first published study of behavioural and psychiatric characteristics of children with Noonan's syndrome. Although there are no agreed criteria for defining a behavioural phenotype of a given condition,¹³ our results highlight certain characteristics that were present in greater than 50% of a sample of children with definite Noonan's syndrome. They suggest a behavioural profile for Noonan's syndrome. Stubbornness, clumsiness, mood problems, communication problems, and faddy eating were described for more than half of our sample. Notably few children showed autistic features, sleep difficulties, severe aggression, sexualised behaviours, or anxiety. These findings are in agreement with the pilot study of Hill.¹⁰ Results from the measure of parent reported psychiatric symptomatology agree with these with these findings, outlining problems on two scales of the CBCL; social relationships and attention deficit. For the remaining six scales of the CBCL (withdrawn, somatic complaints, anxious/depressed, thought problems, delinquent behaviour, aggressive behaviour) few problems are reported. This is a small sample and, as yet, there are no comparison data for the BPQ.

A number of factors other than genotype will be contributing to any describable patterns of behaviour of children with Noonan's syndrome such as parenting styles and psychosocial stresses within the family. Noonan's syndrome is a disorder with important medical implications and the diagnosis may have a significant psychological impact on families. Receiving a diagnosis of Noonan's syndrome may influence parent-child relationships and, secondarily, behaviour. Studying non-affected siblings within the same family has been used as a way of controlling for intrafamilial sources of behavioural symptoms. We found a higher rate of parent reported behavioural problems in children with the syndrome compared with sibling controls. In our small study, this difference failed to reach statistical significance. We note that the rate of 'caseness' on the CBCL in the Noonan's syndrome group was nearly 50% whereas in general population studies, the rate is usually in the order of 15%.¹⁴ The psychiatric symptoms described for children with Noonan's syndrome are non-specific and likely to have complex multiple setiological factors. This finding may highlight a need for specialist provision for some families. We hope that this knowledge about the behavioural and psychiatric aspects of Noonan's syndrome may contribute to diagnosis and subsequent management of children and families with as well as acting as core data for comparison with other genetic syndromes.

We wish to thank secretarial staff in the department of genetics, staff nurse Carolyn Lathan, and all of the children and families who participated in our study.

Full results of the BPQ and CBCL are available on request from the first author.

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