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Social functioning in adults with neurofibromatosis type 1

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ABSTRACT

Neurofibromatosis type 1 (NF1) is a common single-gene disorder characterised by a diverse range of cutaneous, neurological and neoplastic manifestations. It is well recognised that children with NF1 have poor peer interactions and are at risk for deficits in social skills. Few studies, however, have examined social functioning in adults with NF1. We aimed to determine whether adults with NF1 are at greater risk for impairment in social skills and to identify potential risk factors for social skills deficits. We evaluated social skills in 62 adults with NF1 and 39 controls using self-report and observer-report measures of social behaviour. We demonstrate that adults with NF1 exhibit significantly less prosocial behaviour than controls. This deficit was associated with social processing abilities and was more evident in males. The frequency of antisocial behaviour was comparable between the two groups, however was significantly associated with behavioural regulation in the NF1 group. These findings suggest that poor social skills in individuals with NF1 are due to deficits in prosocial behaviour, rather than an increase in antisocial behaviour. This will aid the design of interventions aimed at improving social skills in individuals with NF1.

1. Introduction

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder with an estimated frequency of 1 in 2700 births that occurs equally across genders and ethnic groups (Evans et al., 2010). The most common clinical manifestations involve the skin and include café au lait spots, axillary and groin freckling and cutaneous peripheral neurofibromas. Common complications include central nervous system tumours, scoliosis, and plexiform neurofibromas (Korf, 2002). There is considerable variation in the clinical manifestations of NF1, even within the same family, with some individuals displaying a mild phenotype and others with more severe physical complications. Cognitive deficits are the most common feature of NF1 but are also highly variable between individuals. Intellectual impairment is present in 6–7% of patients (Ferner, Hughes, & Weinman, 1996; Hyman, Shores, & North, 2005), 30–50% satisfy the diagnostic criteria for attention deficit hyperactivity disorder (ADHD) (Hyman et al., 2005; Kayl, Moore, Slopis, Jackson, & Leeds, 2000; Mautner, Kluwe, Thakker, & Leark, 2002; Pride, Payne, & North, 2012), and up to 80% have either learning disability or deficits in executive or visuospatial function (Hofman, Harris, Bryan, & Denckla, 1994; Hyman, Shores, & North, 2006; Hyman et al., 2005; Krab et al., 2008; Pavol et al., 2006).

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While a considerable amount of research has focused on the cognitive sequelae of NF1, the psychosocial impact of the disorder has received less attention. Studies of social functioning have typically employed broader behavioural ratings such as the Child Behaviour Checklist (CBCL) and demonstrate that children with NF1 have significantly more social problems (such as loneliness, clinginess, immaturity) than unaffected children (Dilts et al., 1996; Johnson, Saal, Lovell, & Schorry, 1999; Noll et al., 2007). Social skills have been measured more directly in children with NF1 with mixed results. Noll et al. (2007) assessed social behaviour and peer acceptance in children with NF1 and unaffected peers using teacher, peer and self-report on the Revised Class Play instrument. While children with NF1 did not have lower self-ratings, they were perceived by teachers and peers as displaying less leadership behaviour, as more sensitive and isolated, and less well liked than comparison children. In contrast, Barton and North (2004) found no difference in social skills between children with NF1 when compared to unaffected siblings (measured by the Social Skills Rating System) (Barton & North, 2004). There is evidence for elevated symptoms of autism spectrum disorder (ASD) in children with NF1, with recent evidence suggesting that 14–29% of children with NF1 display severe levels of symptomatology that have been associated with a diagnosis of ASD (Garg et al., 2012; Walsh et al., 2012).

Despite significant social problems in children with NF1 and evidence that adults with the condition frequently struggle to maintain interpersonal relationships (Ablon, 1996), research into the nature of social functioning in adults with NF1 is limited. The burden of NF1 on social functioning has been highlighted by interviews of adult patients (Ablon, 1996; Benjamin et al., 1993; Hummelvoll & Antonsen, 2013; Samuelsson & Riccardi, 1989); reduced social networks, feelings of loneliness, and more social problems such as social withdrawal, are reported by adults with NF1 as a major concern. Nonetheless, there have been no quantitative studies to date that have examined the social skills of adults with NF1 or the underlying reasons for impaired social functioning. A key issue in this regard is whether adults with NF1 are aware of their deficits; previous studies in children with NF1 have shown that they have limited insight into their social abilities (Barton & North, 2004; Noll et al., 2007). It is also unclear whether comorbidity for cognitive impairment or more severe physical manifestations of the disorder contribute to deficits in social functioning.

On this basis, we sought to compare social skills between a sample of adults with NF1 and a sample of unaffected adults using the Social Performance Survey Schedule (SPSS). We hypothesised that social deficits will be a common feature of adulthood NF1 and that adults with NF1 would exhibit poorer awareness of any deficit in social skills. We also aimed to determine how disease severity, demographic and cognitive variables (executive functions, IQ, social processing) relate to social functioning in adults with NF1.

2. Materials and methods

2.1. Participants

Adults who satisfied the diagnostic criteria for NF1 based on a National Institutes of Health (NIH) statement (NIH, 1988) were identified from Neurogenetic Clinic databases from hospitals within local urban and suburban regions including Royal North Shore Hospital, Westmead Hospital and The Children's Hospital at Westmead. Individuals with NF1 were also recruited via an online advertisement on The Children's Tumour Foundation of Australia website. Medical files were examined to determine eligibility for participation. Exclusion criteria for NF1 patients were as follows: (1) less than 18 years or more than 60 years of age, (2) insufficient understanding of the English language, (3) history of co-existing genetic or neurological condition, (4) abnormal vision or hearing that could not be corrected to normal and (5) unable to complete questionnaires independently. Of the 81 NF1 patients contacted, 14 refused to participate, and five were excluded due to either insufficient language abilities ($n = 2$), severe psychiatric disturbance i.e. psychosis ($n = 1$), or insufficient reading abilities to complete the questionnaire ($n = 2$). Thus, 62 NF1 patients participated in the study.

In order to provide a modern comparison group for this study, we recruited 39 unaffected adults between the ages of 18 and 60 from local urban and suburban areas. Participants were recruited from a variety of sources: (1) normal individuals recruited from the wider community via advertisements ($n = 22$), (2) parents of unaffected children who were controls in a concurrent study examining the cognitive phenotype of toddlers with NF1 ($n = 15$), (3) parents of children with sporadic NF1 (non-familial) who were seen at the hospital's Neurogenetic Clinic ($n = 2$). Control participants were excluded from the study based on the same exclusion criteria used for NF1 patients. Control participants with a reported psychiatric history (e.g. ASD, ADHD, psychiatric disturbance) were additionally excluded.

2.2. NF1 disease severity

Clinical severity of NF1 was rated with Riccardi's severity scale (Riccardi & Kleiner, 1977). Grade 1 indicates that few features of NF1 are present with no compromise of health (e.g. café au lait spots and freckling only). Grade 2, or mild NF1, indicates physical signs of NF1 that are obvious to others and are a concern, but without significant compromise of health. Grade 3, or moderate NF1, indicates a significant compromise of health but symptoms can be managed reasonably well. Finally, Grade 4, or severe NF1, indicates the presence of serious compromise that is intractable, is managed or treated only with difficulty and is associated with a shortened life span. Severity was assigned retrospectively from patient medical files and interview transcripts. One investigator (N.P.) and a clinical geneticist (both who have extensive experience with the clinical management of NF1 patients), rated severity in 15 of the same NF1 participants to establish inter-rater reliability.