

An Assessment of Parent Treatment Needs for Children with Neurofibromatosis Type 1

Heather L. Thompson, M.Sc., University of Utah

Sara Hunt, M.S., Utah State University

April 25, 2008

Overview of Project: Two focus groups were conducted with parents/caregivers of children with NF1 to ascertain their need for services beyond standard medical care. The focus groups were facilitated by a speech and language pathology graduate student and a psychology graduate student. Parent/caregiver responses suggested the need for a multi-disciplinary NF1 clinic to better serve these children and their families.

Introduction

Neurofibromatosis Type 1 (NF1) is an autosomal dominant neurocutaneous disorder. NF1 has a prevalence of approximately 1/3000-4000 (Friedman, 1999; Stumpf et al, 1988). NF1 is congenital, that is, it is present at birth. Children with NF1 have a number of concerns which may negatively affect central nervous, ocular, skeletal and epidermal system function (Ferner et al, 2006; Lee & Stevenson, 2007; Stevenson et al, 2007; Young, Hyman & North, 2002). Physical findings of the disorder may or may not include the presence of café au lait macules, lisch nodules, plexiform or neurocutaneous neurofibromas, or tibial bowing, which result from deletions in three regions of the TBR gene. Approximately 98% of patients diagnosed with NF1 have more than six café-au-lait macules (Korf, 1992). Deletions are located on the long arm (q11.2) of Chromosome 17 (Viskochil et al, 1990; Viskochil, 1999).

A number of behavioral characteristics have been reported for individuals with NF1. For example, patients have been reported to have learning disabilities (Coude et al, 2006; Eldridge et al, 1989; Hyman, Shores & North, 2006; North et al, 1994; Ozonoff, 1999), visuo-spatial/visuo-motor impairment (Eldridge et al, 1989; Eliason, 1986; Hofman et al, 1994; Riccardi, 1989; Varnhagen, et al, 1988), attention deficit/hyperactivity disorder (ADHD; Barton & North, 2004, 2006; Eldridge et al, 1989; Hofman et al, 1994; Hyman et al, 2006; Koth et al, 2000; Mautner et al, 2002), hearing impairment (Hallpike, 1963; Poissant, Lustig & Jackler, 1996), lower reading, low spelling and/or verbal abilities (Eliason, 1986; Hofman et al, 1994; Hyman et al, 2005; Stine & Adams, 1989), cognitive delay (Hyman et al, 2005), voice concerns (Alves de Rezende, 2007) and social-emotional problems (Johnson et al, 1999). Resonance concerns have been found in this population (Ferner et al, 1996; Lorch et al, 1999; North et al, 1995). Resonance concerns include the inability to achieve closure of the velopharyngeal port (i.e., coupling of the oral and nasal cavities) during the production of oral speech sounds (e.g., “p”, “f” & “s”).

While it is understood that children with NF1 require frequent visits to a pediatrician for the purpose of monitoring development, it is unknown which other allied services may be required and beneficial in the early stages of development for preschool children with NF1. As speech and language delays have been observed in this population of children (Thompson, et al, 2008) and there is some indication of possible psychological concerns, other services may be needed as part of team care for these children. Therefore, the primary objective of the proposed

study is to determine the services which will be of greatest assistance to young children with NF1. To this end, the following questions were addressed:

1. What services do children with NF1 currently receive?
2. What do parents of children with NF1 report needing in terms of services other than medical treatment?

Method

Participants

Parents of eighteen preschool children who receive care through Primary Children's Medical Center in Salt Lake City (UT) and who either had a formal diagnosis of NF1 or 6 or more café au lait macules, were invited to participate. The invited study population included males and females as well as individuals from different racial, ethnic and socio-economic groups.

Parents were mailed a letter informing them of the purpose and procedures of the study. The letter indicated that they would receive a telephone call from one of the researchers (Heather Thompson) within ten days of receiving the letter. The letter indicated that if they did not want to receive this phone call that they could call and leave a message indicating so. No telephone calls were received indicating that any participants did not want to participate.

Eighteen families were telephoned regarding the study. During the telephone call, participants were informed of the purpose, procedures and time requirements of the study. They were told that the study consists of one meeting, based on the schedule and the convenience of the participants. Parents were also informed that they would be paid \$25 for their involvement in the focus group.

Out of the eighteen families, four parents/caregivers participated in the study. Table 1 provides more information about the participants and their children. All names have been changed to maintain privacy.

Table 1
Participant Profiles

Parent Name (Child Name)	Child's Age	Background
Mary (Jonathon)	3	NF1 suspected at 2 years of age; no family history; diagnosed October 2007
Ron and Barb (Kristin)	5	Diagnosed one month ago; no family history
Jane (Grandsons: Chris Brad)	5 3	Diagnosed at birth; family history of NF 1 on father's side

Procedures

Participants met at the University of Utah for one of two 1-hour focus group sessions. Prior to the start of the focus group, investigators obtained informed consent (documented by a signed consent form). Following obtaining informed consent, parents were asked a series of questions (Appendix A). Health care professionals from the areas of speech-language pathology, genetic counseling, psychology, rehabilitation, pediatric dentistry and business administration

attended the focus groups for the purpose of providing information to parents regarding possible services available to them.

Video and audio-recordings were obtained using a Sony Electret Condenser microphone (ECM-55B) connected to a Marantz portable cassette recorder (PMD430).

Results

The audio recordings were transcribed and coded for analyses. Transcriptions were reviewed by two investigators in order to ensure reliability of the data. Responses were coded according to the two principle areas of interest: 1) services currently used by children with NF1 and 2) services parents of children with NF1 would like to access as part of a multi-disciplinary clinic. Each area of interest was then analyzed for themes or patterns.

Current Services Used

Regular appointments. All of the parents/caregivers reported their child had been seen by a geneticist in addition to receiving yearly eye exams.

Additional services. Only Jane's grandsons had accessed early intervention services including speech/language therapy. Neither Jonathon nor Kristin had ever received speech/language services. It should be noted that the four children reported on in this study had all participated in a research project exploring the speech and language functioning of pre-school aged children with NF1 (Thompson, et al., 2008). Testing results from that study indicated that Kristin and Brad demonstrated a need for speech/language therapy. Jane also reported Chris is being tested by his school for ADHD.

Requested Services

Health care providers. All of the parents/caregivers identified a need for ongoing assessment and treatment for possible speech/language problems. They also expressed a shared interest in mental health services for their children to address a number of possible needs:

Mary: I would imagine that for him, I mean it is a very physical [condition]. If it developed to that point where you know people could tell from looking at him, I'm sure that could be hard so having access to that and maybe for the whole family.

Ron and Barb and Jane also indicated it would be helpful to have access to a mental health professional to evaluate and treat children with NF1 for learning disabilities and ADHD.

The families also reported a need to have orthopedists, neurologists, dermatologists, and oncologists included in their child's health team in addition to geneticists and optometrists.

Parent support. Mary, Ron and Barb all suggested a centralized place to inform parents about the symptoms and possible outcomes of children with NF1 as well as to serve as a support system between parents. Barb stated, "I wish as a parent they had a class because I do not like the unknown, I do better if I know."

Child support. Mary recommended a support group be established for youth with NF1 so her son could connect with other children with the same diagnosis. She said, "This population is not super huge so finding them in your neighborhood is not going to happen." The other families also expressed an interest in wanting their children to communicate with other youth with NF1.

Discussion

Common themes were identified in the responses from the three families who participated in the focus groups. Geneticists and optometrists were reported to be part of the standard care received by all of these children. Similar to results from previous studies, some of the children reported on in this study required evaluation and treatment for speech/language delays and for concerns of ADHD.

When asked to identify additional care needs for their children with NF1, all of the families indicated the need for ongoing speech/language services, mental health professionals, and various medical providers (e.g., orthopedics, neurology). Children in two of the families who participated had been diagnosed within the past six months and these parents expressed a desire to have more education about NF1 as well as a parent support group to share experiences with other families. It was also suggested that youth with NF1 may also benefit from a support system to connect them with each other. These responses indicate a multi-disciplinary clinic could better meet the needs of these children and families, and that future research is needed to understand more clearly the speech and language and psychological issues of youth with NF1.

References

- Alves de Rezende, N. (2007). High incidence of voice and oral motor control disturbances in NF1: a preliminary report, Poster Session, The Children's Tumor Foundation NF Conference, June 10-12th, Park City, UT.
- Barton, B. & North, K. (2004). Social skills of children with neurofibromatosis 1. *Developmental Medicine & Child Neurology*, 46, 553-563.
- Coude, F.X., Mignot, C., Lyonnet, S. & Munnich, A. (2006). Academic impairment is the most frequent complication of neurofibromatosis type-1 (NF1) in children, *Behavior Genetics*, 36(5), 660-664.
- Eldridge, R., Denckla, M.B., Bien, E., Myers, S., Kaiser-Kupfer, M., Pikus, A., Schlesinger, S., Parry, D., Dambrosia, J., Zasloff, M., Mulvihill, J. (1989). Neurofibromatosis type 1 (recklinghausen's disease), *ADJC*, 143, 833-837.
- Eliason, M.J. (1986), Neurofibromatosis: Implications for learning and behavior, *Journal of Developmental and Behavioral Pediatrics*, 7, 175-179.
- Ferner, R.E., Huson, S.M., Thomas, N., Moss, C., Willshaw, H., Evans, D.G., Upadhyaya, M., Towers, R., Gleeson, M., Steiger, C., Kirby, A. (2006). Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. *J Med Gen.*, 44, 81-88.
- Ferner, R.E., Hughes, R.A.C., Wenman, J. (1996). Intellectual impairment in neurofibromatosis 1. *Journal of Neurological Sciences*, 138: 125-133.
- Friedman, J.M. (1999). Epidemiology of Neurofibromatosis Type 1. *American Journal of Medical Genetics*, 89, 1-6.

Hallpike, C.S., (1963). Stapes fixation by an intra-labyrinthine 'seedling' neurofibroma as a cause of conductive deafness in a case of von Recklinghausen's disease, *Acta Otolaryngologica Supplementum*, 183, 62-65.

Hofman, K.J., Harris, E.L., Bryan, R.N. & Denckla, M.B. (1994). Neurofibromatosis type 1: The cognitive phenotype. *The Journal of Pediatrics*, 124, S1-S8.

Hyman, S.L., Shores, A., & North, K.N. (2005). The nature and frequency of cognitive deficits in children with neurofibromatosis type 1, *Neurology*, 65: 1037-1044.

Hyman, S.L., Shores, A., & North, K.N. (2006). Learning disabilities in children with neurofibromatosis type 1: subtypes, cognitive profile, and attention-deficit-hyperactivity disorder. *Developmental Medicine & Child Neurology*, 48, 973-977.

Johnson, N.S., Saal, H.M. Lovell, A.M., Schorry, E.K. (1999). Social and emotional problems in children with neurofibromatosis type 1: Evidence and proposed interventions, *The Journal of Pediatrics*, 134, 767-772.

Koth, C.W., Cutting, L.E., Denckla, M.B. (2000). The association of neurofibromatosis type 1 and attention deficit hyperactivity disorder, *Child Neuropsychology*, 6, 185-194.

Korf, B.R., (1992). Diagnostic outcome in children with multiple café au lait spots. *Pediatrics*, 90(6), 924-7.

Lee, M. & Stevenson, D.A. (2007). Recent developments in neurofibromatosis type 1, *Current opinion in neurology* 20, 135-141.

Lorch, M., Ferner, R., Golding, J., Whurr, R. (1999). The nature of speech and language impairment in adults with neurofibromatosis 1, *Journal of Neurolinguistics*, 12:157-165.

Mautner, V.F., Kluwe, L., Thakker, S.D., Lark, R.A., (2002). Treatment of ADHD in neurofibromatosis type 1. *Developmental Medicine & Child Neurology*, 44, 164-170.

North, K., Joy, P., Yuille, D., Cocks, N., Mobbs, E., Hutchins, P., McHugh, K., de Silva, M (1994). Specific learning disability in children with neurofibromatosis type 1: significance of MRI abnormalities, *Neurology*, 44, 878-883.

Ozonoff, S. (1999). Cognitive impairment in neurofibromatosis 1. *American Journal of Medical Genetics*, 89, 45-52.

Poissant, S.F., Megerian, C.A. & Hume, D. (2003). Cochlear implantation in a patient with neurofibromatosis type 1 and a profound hearing loss: Evidence to support a cochlear site of lesion, *Otology and Neurology*, 24, 751-756.

Riccardi, V.M. Neurofibromatosis, natural history, and pathogenesis. 1992. Baltimore: Johns Hopkins University Press.

Stevenson, D.A., Loyer-Mileur, L.J., Murray, M., Slater, H., Shang, X., Carey, J.C., Dube, B., Viskochil, D.H. (2007). Bone mineral density in children and adolescents with neurofibromatosis type 1. *Journal of Pediatrics*, Jan, 83-88.

Stine, S.B., & Adams, W.V. (1989). Learning problems in Neurofibromatosis patients. *Clin Orthop.*, 245, 43-48.

Stumpf, D.A., Alksne, J.F., Annegers, J.F., Brown, S.S., Conneally, P.M., Housman, D., et al, (1988). Neurofibromatosis conference statement national institutes of health consensus development conference, *Arch Neurol*, 1988; 45:575-8.

Thompson, H., Chapman, K., Viskochil, D & Stevenson, D. Speech and Language Development of Preschool Children with Neurofibromatosis 1 [Abstract] Presentation at the Children's Tumor Foundation Annual Convention, Naples, FL 2008.

Varnhagen, C.K., Lewin, S., Das, J.P., Bowen, P., Ma, K., Klimek, M. (1986). Neurofibromatosis and psychological processes. *J Dev Behav Pediatr.*, 9, 257-65.

Viskochil, D. (1999). Neurofibromatosis 1. *American Journal of Medical Genetics*, 89, v-vii.

Viskochil, D., Buchberg, A.M., Xu., Cawthorn, R.M., Stevens, J., Wolff, R.K., Culver, M., Carey, J.C., Copeland, N.G., Jenkins, N.A., White, R., O'Connell, P. (1990). Deletions and a translocation interrupt a cloned gene at the Neurofibromatosis Type 1 locus. *Cell*, 62, 187-192.

Young, H., Hyman, S. & North, K. (2002). Neurofibromatosis 1: Clinical review and exceptions to the rules, *Journal of Child Neurology*, 17, 613-626.

Appendix A
Questions Posed During the Focus Groups

1. What is the age of your child?
2. What is your child's biological sex?
3. What educational services does your child currently receive?
4. What medical services does your child currently receive?
5. Does your child receive all medical services through Primary Children's Medical Center?
6. Does your child currently receive speech-language services or early intervention?
 - a. If yes, does your child receive these services through his/her:
 - i. School
 - ii. Daycare
 - iii. Other
 - iv. Privately
 - b. If no, do you feel that your child may benefit from receiving speech-language services?
7. What services would you like to have included as part of an interdisciplinary team care for children with Neurofibromatosis 1?