

Children with Disabilities are Often Misdiagnosed Initially and Children with Neuropsychiatric Disorders are Referred to Adequate Resources 30 Months Later Than Children with Other Disabilities

Alli-Marie Tuominen-Eriksson · Yvonne Svensson · Ronny K. Gunnarsson

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Abstract Disabilities in a child may lead to low self-esteem and social problems. The lives of parents and siblings are also affected. Early intervention may decrease these consequences. To promote early intervention early referral to adequate resources is essential. In a longitudinal retrospective observational study it was found that children with neuropsychiatric disorders without mental retardation were referred 30 months later than other children. Agreement between the referrer's identification of the main disability and the habilitation center's was low with Kappa coefficient 0.44. Whereby agreement on diagnosis between referrer and habilitation centers was low, earlier referral should be promoted.

Keywords Asperger syndrome · Attention deficit disorder with hyperactivity · Autistic disorder · Early diagnoses · Referral and consultation · Mental retardation

Introduction

In a Swedish survey from 1987, 8.4 % of all children were identified as presenting with a chronic illness or disability

(Westbom and Kornfält 1987). Most common were mild or moderate conditions but 0.66 % of all children had a severe disability, usually caused by mental and nervous system disorders and congenital malformations (Westbom and Kornfält 1987).

Neuropsychiatric disorders such as attention-deficit/hyperactivity disorder (ADHD), Asperger's syndrome and childhood autism have received increased attention revealing a prevalence of 2–18 % for all children (Rowland et al. 2002).

The majority of mental and nervous disorders and congenital malformations in children are present at birth or acquired during the first year of life. These disorders may present as movement disabilities, speech- and language disabilities, neuropsychiatric disabilities, mental retardation, hearing or visual impairment. In many countries, including Sweden, disabilities are classified and diagnosed according to the International Classification of Diseases version 10 (ICD-10). ICD-10 was endorsed by the forty-third world health assembly in May 1990 and came into use in WHO member states as from 1994.

These impairments may lead to low self-esteem, social problems such as forming and maintaining relationships, completing an education and becoming economically self sufficient (Pratt et al. 1999; Klassen et al. 2004; Rehm and Bradley 2006). Furthermore, affected children often have more medical problems than children without these disabilities (Lin et al. 2006). Having a disabled child will also affect the lives of parents and siblings (Ray 2002; Klassen et al. 2004). Measures provided for the extra needs of these children and their families (Shonkoff et al. 1992) result in increased costs for society.

In Sweden, at the time of this study, all children were screened for developmental disorders at birth, when discharged from the maternity ward, within screening

A.-M. Tuominen-Eriksson (✉) · Y. Svensson
Child and Adolescent Habilitation Center in Fyrbodal County,
Barnhabiliteringen Mariedal, Restadvägen 24,
462 54 Vänersborg, Sweden
e-mail: alli-marie.t-eriksson@vgregion.se

R. K. Gunnarsson
Department of Public Health and Community Medicine,
University of Gothenburg, Gothenburg, Sweden

R. K. Gunnarsson
Cairns Clinical School, School of Medicine and Dentistry, James
Cook University, Cairns, Australia

programs at a child health care center (CHC) and school, for children aged 7–16. At the CHC the child was routinely examined by a primary care physician or pediatrician at 2, 6, 10–12 months and 5 years of age. The child also met a nurse at the CHC when aged 1 month, 1.5 years, 2.5–3 years and 4 years. The children met a school nurse at the age of 6–7, 10 and 13–14 years. Screening at birth, discharge from the maternity ward and the CHC consisted of a structured set of tests while the screening at school consisted mainly of a discussion of possible social and learning problems, drug abuse, and physical activity. If potential problems were identified the child may have been referred to a pediatric clinic or habilitation center for children with disabilities. As soon as a disability such as movement disability, mental retardation, acquired brain injury or some neuropsychiatric disorders were discovered the recommendation was for the child to be referred to a habilitation center for children with disabilities. Thus, a child first referred to a pediatric clinic may have later been referred to a habilitation center for children with disabilities.

Child and adolescent habilitation services in Sweden are offered to families with children with a disability such as movement disability, mental retardation, acquired brain injury and some neuropsychiatric disorders, mainly autism and autistic like conditions such as Asperger's syndrome. These services are provided by the National Health Service at regional habilitation centers providing in-depth assessment and support. The staffs at these centers consist of physiotherapists, occupational therapists, speech and language pathologists, psychologists, social workers, special education teachers. Nurses are present in many of these centers. Pediatric neurologists were previously often a part of the team but in recent years they often join the team at regular intervals as consultant physician.

Although not clearly proven for many conditions (Cunningham 2004), early intervention is likely to decrease the consequences of disability (Shonkoff and Hauser-Cram 1987; Majnemer 1998; Bailey et al. 2004, 2005; Feldman et al. 2008). However, early intervention requires early detection. Fortunately, many disabilities can often be detected early (Valtonen et al. 2004).

A severe disability in a child is usually observed very early by parents (at 7–8 months) and at child health care centers 1.4 months later (Bailey et al. 2004). However, referral to initiate intervention can be further delayed another 5.2 months after detection (Bailey et al. 2004). Mild or moderate disorders such as ADHD may be detected as early as 4 years of age (Valtonen et al. 2004). However, it seems that this and similar disabilities are identified much later (Lock et al. 1986).

To promote early referral, the findings of Lock et al. (1986) should be confirmed from a country other than the

USA. It is also of interest to clarify whether the child's main disability remains concealed until referral and subsequent careful investigation at the habilitation center, or if the referrer has already guessed correctly. One aim of this study was to investigate if the referrer correctly identified the child's main disability. Another aim was to see if a child's age at referral to a habilitation center depended on the type of disability as diagnosed by the habilitation center.

Methods

All consecutive referrals from January 1, 1998 to December 31, 2001 to a habilitation center for children with disabilities in the south west region of Sweden, a mixture of urban and rural population, were analyzed. The Ethical Committee Göteborg University approved the study.

The referrers' preliminary diagnoses and the habilitation center's final main diagnoses were both merged into six diagnostic groups; movement disorders (MD), language disorders without mental retardation (LD), neuropsychiatric diagnosis with mental retardation (NP + MR), neuropsychiatric diagnosis without mental retardation (NP), mental retardation (MR) and finally miscellaneous diseases (Misc). MD in this context refers to children in need of physiotherapy. For children having a final main diagnosis within group MD, the underlying cause was in 55 % diseases of the nervous system. The Misc group consisted of juvenile arthritis, acquired brain injury and birth trauma.

Selection of Participants

The center participating in this study offered child and adolescent habilitation services as described in the introduction. It served a geographical target area that during 1998–2001 had a mean of 148 272 inhabitants, 33,430 of these were children aged ≤ 17 years. During these years an average of 1,396 children were born annually. The longest travel to the habilitation center would be 100 km.

Beside the diagnostic panorama for all centers in Sweden, at the time for data collection this habilitation center also served children with severe speech and language disorders and severe ADHD. All consecutive referrals to the center from 1998 to 2001 were included. During these years children could be referred due to an unspecified developmental delay without including an established medical diagnosis. Most referrals were from CHC, pediatric clinics, school health care, a child psychiatric clinic and primary health care centers. Some referrals came from other habilitation centers as a result of the child moving into the target area.

Measurements

Referrals and medical records in the habilitation center written by physicians were scanned and information regarding presence or absence of speech and language delay and presence of epilepsy was documented. Children could have had mild or moderately delayed speech and language ability without obtaining a final main diagnosis within diagnostic group language disorders without mental retardation. These children had other disabilities and thus obtained a main final diagnosis within another diagnostic group. Information regarding languages spoken in the child's immediate environment, and whether the child was a twin or a triplet was also noted. The referrer's diagnosis, children's sex and age at referral and final main diagnosis from the habilitation center were registered. The latter was determined after careful investigation and usually not provided until 1–2 years after referral.

All incoming referrals were first discussed in a referral team with representatives from the different disciplines in a habilitation team. When the referral was accepted it was passed on to the habilitation team consisting of a pediatric neurologist, a physiotherapist, an occupational therapist, a speech and language pathologist, a psychologist, a social worker, a special education teacher and a nurse. The team planned all further assessments depending on suspected types of disabilities. The diagnostic procedures often included physical examination, basic laboratory testing, electroencephalogram, chromosomal analysis, magnetic resonance imaging of brain and auditory brainstem response audiometry. Psychological testing and evaluation by other disciplines in the team was done. Depending on preliminary findings the assessment plan could be revised. A final chief diagnosis set by the habilitation center was then merged into the six diagnostic groups; MD, LD, NP + MR, NP, MR and finally Misc.

Results

During 1998–2001, 293 children and adolescents were referred to the habilitation center. The numbers referred in a year varied from 66 to 83 corresponding to 0.04–0.06 % of the area's total population and 0.20–0.25 % of child population aged ≤ 17 years. Twenty-nine of the 293 referrals (9.9 %) came from an outside habilitation center due to children moving into the area. The mean proportion of boys for all 4 years was 68.8 % with a range of 63.5–77.6 %. Eliminating 29 children referred by other habilitation centers yielded a mean age at referral of 73.5 months (median 71 months, standard deviation 53.3 months, interquartile range 31–106 months, range 0–203 months) with no difference between girls and boys. There was no trend during the 4 year period toward an

increase or decrease either in number of referred children, proportion of boys or age at referral. Nine (3.1 %) were twins and 1 (0.3 %) was a triplet. Thirty-two (10.9 %) children had more than one language spoken in their immediate environment. 135 children (46 %) had delayed speech and language ability.

A final main diagnosis was set by the habilitation center in 257 of 293 children. Of the 36 children not obtaining a final main diagnosis, 21 had birth injuries they recovered from within a year after referral (12 injuries to the peripheral nervous system and 9 sternomastoid injuries). Two children were ill at referral but recovered within a year after referral (1 child with an acquired brain injury and 1 referred for sequels of encephalitis) and for another 13 children referred for a final opinion by the habilitation center, it was determined that the children had no disability.

Association Between Referrer's and Habilitation Center's Diagnosis

The proportions of different problems preliminarily identified by the referrer as the child's main disability did not change over time. Information about preliminary problem identification by referrer and final main diagnosis by the habilitation center was obtained in 233 children. The association between all referrer's problem identification (other habilitation centers excluded) and final main diagnosis by the habilitation center was low, with Kappa coefficient 0.44 (95 % confidence interval of Kappa coefficient 0.35–0.52) (Table 1).

The association between problem stated in the referral and final main diagnosis by habilitation center was much

Table 1 Association between referrer and habilitation center concerning the child's main disability

		Main disorder by habilitation center					
		MD	LD	NP + MR	NP	MR	Total
Main problem by referrer	MD	35	2	1	0	7	45
	LD	1	16	4	4	14	39
	NP + MR	2	3	12	5	5	27
	NP	1	5	5	25	3	39
	MR	6	7	7	3	31	54
	Total	45	33	29	37	60	204

29 referrals from other habilitation centers when patients move into area are excluded. Furthermore, 24 referrals classified in the group of Miscellaneous disorders are omitted

The bold values illustrate that referrer and habilitation center agree on the main disability

MD movement disorders, *LD* language disorders without mental retardation, *NP + MR* neuropsychiatric diagnosis with mental retardation, *NP* neuropsychiatric diagnosis without mental retardation, *MR* mental retardation

higher for the 29 children referred by another habilitation center, with a Kappa coefficient of 0.95 (95 % confidence interval of Kappa coefficient 0.85–1.00).

Age at Referral

Eliminating 29 children referred by other habilitation centers yields a referral age for diagnostic group MD of 44 months (Standard deviation = SD 1, 5), group LD 84 months (SD 33.0), group NP + MR 72 months (SD 44.0), group NP 101 months (SD 46.1) and for group MR 71 months (SD 47.1). Age at referral differed between diagnostic groups ($p < 0.0001$, ANOVA). Children in group NP were referred later than other children ($p = 0.0001$, Student's t test). Children with delayed speech and language ability were referred at 64 months of age compared to 85 months for children without this delay ($p = 0.0059$, due to unequal variances found by Bartlett's test for inequality of population variances Kruskal–Wallis test for two groups were used).

A multiple linear regression analysis was performed to further clarify the association between age at referral (dependent variable) and diagnostic group (independent binary class variable) while compensating for other confounding factors such as sex, multiple languages spoken at home, born twin or triplet and presence of epilepsy. One regression was made for each diagnostic group. Referrals from other habilitation centers were excluded because children moving into the target area were almost always diagnosed at a previous habilitation center several years earlier. In this multiple regression model children with movement disorders (group MD) were referred 35.6 months earlier than other children ($p = 0.03$) and children with neuropsychiatric diagnosis without simultaneous mental retardation (group NP) were referred 30.0 months later than other children ($p = 0.0007$).

Discussion

The association between the referrer's problem identification and final main diagnosis by the habilitation center was lower than expected, with Kappa coefficient 0.44. This supports the need for promoting earlier referral. Furthermore, this study showed that children with movement disorders were referred 36.6 months earlier than other referred children. Children with neuropsychiatric diagnosis without mental retardation were referred 30.0 months later.

Methodological Aspects

In choosing between analyzing a sample of referrals from several habilitation centers or all referrals from one center we chose the latter. The main reason was that it simplified

retrieval of data. The disadvantages of doing so were judged minimal whereby most habilitation centers in Sweden have similar target populations with similar goals, staff and financial resources. Thus, the generalizability of the results to other settings in Sweden should be acceptable. The main finding that children with neuropsychiatric diagnoses without mental retardation were identified and given optimal support later than what is considered adequate is probably true of most countries.

Evaluating the nature of how referrers set a diagnosis and the factors in the habilitation center leading to an altered diagnosis could also have been accomplished using an empiric-holistic (qualitative) approach. Due to practical reasons, it was not possible to include this interesting approach in this study. However, this could be a suitable approach for future studies.

It is difficult to evaluate the presence of mild mental retardation in very young children. The youngest group was children with movement disorders. Mental retardation may be prevalent among them, especially in those with diseases of the nervous system having the diagnostic code ICD-10 G80 (cerebral palsy). However, these children were maintained in the group movement disorders as this was the first obvious symptom.

The diagnoses of language disorders without mental retardation (LD) were given to children with severe speech and language impairment with no other diagnosis. Their total intelligence quotient (TIQ) (verbal and performance combined) was normal when tested. However, some children with the diagnosis of language impairment in early childhood received the diagnosis of mental retardation a few years later (Botting 2005). Many children with other diagnoses also have difficulties with speech and language. Therefore we defined the variable "delayed speech and language ability". All children in group LD except two with the diagnosis of specific developmental disorders of scholastic skills (ICD-10 F81) were coded as presenting with delayed speech and language ability. These two children were later diagnosed by speech and language pathologists as having a language disorder.

Since the time of data collection two major changes has taken place. One major change is a shift in focus from support and training in the habilitation center to community based training. In the latter model staff at the habilitation center is a resource and support for parents, local teachers and special education teachers. This change mainly influence what happens after diagnosis and should not have any direct effect on the results of this study. Another major change is that at the time for data collection children being suspected to have a disability were allowed to be referred. At present, all habilitation centers in Sweden require referrals to include a formal diagnosis within one of the above mentioned diagnostic groups. Thus, in some cases,

this entails a further delay whereby the child must await testing and setting of the diagnosis before referral. The consequences might be that delay in establishing a correct diagnosis has increased compared to when data collection was made. Since this study showed that initial preliminary diagnosis was not always correct a further delay may have negative consequences for the child at its family.

In the present study children with the final diagnosis autism were all also mentally retarded.

Movement Disorders

Children with a motor disability received intervention at a much younger age than other referred children. A motor disability, such as cerebral palsy or a muscular disorder does not demand a psychological investigation. Muscle tone and the movement patterns are clearly aberrant and the pediatrician can refer the child to the habilitation center. Since this problem is easily identified early, delayed detection does not appear to be a major problem.

Neuropsychiatric Diagnoses Without Mental Retardation

Children with normal intelligence but with a neuropsychiatric diagnosis were referred 30 months later than children with other diagnoses. This finding is not surprising and confirms the previous findings of Lock et al. (1986). Roberts et al. (2008), who only studied preterm children with disabilities, found similarly that milder disabilities were associated with longer delay to habilitation services. A diagnosis such as Asperger's syndrome or ADHD is a behavioral diagnosis and the child might show different behavior in different situations or environments. Pediatricians can easily observe motor movement but not as easily behavior. Thus, psychological assessment is usually needed, which slows the process of determining diagnosis.

Although the finding that children with normal intelligence but with a neuropsychiatric diagnosis were referred much later than children with other diagnoses is not new, its importance is strongly emphasized by the new finding in this study that agreement on diagnoses between referrers and habilitation centers was poor. This implies that children might not get appropriate support until a referral leads to an adequate diagnosis.

A consequence of late detection and initiation of adequate support is that children may experience many failures, which causes low self-esteem. Learning problems in school may lead to disinterest in school work, and a high risk of anti-social behavior. Thus, delayed detection or referral of these children can have significant consequences.

Referral for proper evaluation usually requires a multidisciplinary team and should be made earlier than at

present. Increased costs for testing probably far outweighs the long term benefits of a family having earlier contact and support from a habilitation center.

Association Between Referrers' and Center's Diagnosis

The association between the referrer's problem identification and final main diagnosis by the habilitation center was lower than expected, with Kappa coefficient 0.44. In Table 1 there was agreement on 119 of 204 patients. Of the remaining 85 patients two main reasons for disagreement appears. One was that the referrer did not perceive mental retardation being the main diagnosis while the habilitation center did (29/85). The other main reason for disagreement was the reverse; the referrer perceived the main diagnosis was mental retardation while the habilitation center did not (23/85). These two reasons combined add up to 61 % of all disagreements. Thus, mental retardation seems to be a major cause for diagnostic confusion.

Establishing mental retardation requires psychological testing. In case of severe mental retardation this does not pose any major diagnostic problem. However, in mild mental retardation parents might not allow psychological testing to avoid stigmatizing their children. The habilitation center has much more resources and experience than the referrer of dealing with these problems. Thus, they are more likely to be able to sort this out.

If referrer and habilitation center disagree who is right? The high correlation (high kappa coefficient) in main diagnosis between different habilitation centers suggests that the habilitation center was right.

Getting a false main diagnosis as mental retardation or, the reverse missing this diagnosis is likely to have negative consequences for the child and its family. Thus, this study supports the need for promoting earlier referral.

Can Intervention at a Habilitation Center Be Initiated Earlier?

Parents are in need of information about the condition of their child, to feel competent in their role as supportive parents. A multidisciplinary team can design a training program that parents and pre-school teachers can carry out. Through the habilitation center, parents can contact other families and exchange advice. Thus, the information and support the habilitation center gives parents makes daily living easier (Michaud 2004; Bailey et al. 2005). Furthermore, it may enhance future development of the child (Ehrmann Feldman et al. 2005; Duby 2007).

It is recommended that intervention start before the age of 3 years (Bailey et al. 2005). Many children are identified at the CHC center at the age of 2.6 years (Miniscalco et al. 2006). Unfortunately, this study showed that children with

a disability were not referred to the habilitation center until an average age of 5½–6 years. In this study no established medical diagnosis was required before referral. Introducing a demand on established diagnosis before referral, as has been done in Sweden after the completion of this study, would probably introduce a further delay before referral to proper intervention (Silverstein et al. 2006).

The association between the suggested diagnoses of the referrers and the final main diagnoses was lower than expected. Therefore, referral to a habilitation center should not depend upon a diagnosis according to ICD-10. Earlier referral could be made to a habilitation center and be based on a description of the developmental delay or problems, rather than the determination of a diagnosis, which is a time consuming process.

A widening of the group of children getting access to habilitation services would be valuable but is a matter of resources. Such approach would probably decrease suffering and also save resources in the long run.

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