Clinical Topics

Diagnosis of Duchenne muscular dystrophy: experiences of parents of sufferers

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Abstract

Sixty nine parents of boys suffering from Duchenne muscular dystrophy were interviewed at home. The interview explored the parents' experiences at the time of their son's diagnosis. Many families had experienced distressing delays (average 2·5 years) between the time they first became aware of symptoms and the time of the diagnosis. On only 18 occasions were both parents told of the diagnosis together. One third of the parents were "not satisfied" with the way the diagnosis had been communicated. Parents want to know as soon as possible if there is something wrong with their child. They should be told the diagnosis together and in private. Full information should be given and a series of contacts should be arranged.

Introduction

Duchenne muscular dystrophy is a neuromuscular disease that affects boys only and is inherited as an X-linked recessive trait. In two thirds of cases there is a family history of the disorder; the remainder are spontaneous mutations. The child initially has no symptoms. The parents may notice early symptoms from the age of 6 months onwards, but the diagnosis is often delayed until the boy is about 4 or 5 years old.

Duchenne muscular dystrophy is a severe and relentlessly progressive disease. The boys first have difficulty in walking, running, or climbing stairs. They fall frequently and have difficulty in getting back on their feet. They experience progressive weakness and at about the age of of 8 or 10 years need a wheelchair. Over the years they become more disabled; their arms become weaker and eventually the muscles of their faces, hands, and chest are affected. Death usually occurs between the ages of 16 and 25.

Parents of boys with Duchenne muscular dystrophy may be told the diagnosis in various ways and after variable delays. Parents in three parts of Britain were asked to discuss their experiences.

Method

Sixty six families from three separate geographical areas of Britain were asked to take part in the study. In 53 families one or both parents agreed to be interviewed: 37 mothers were interviewed alone and 16 couples were interviewed together. At the time of the interviews 13 boys were aged 4 years; 26 were between 5 years and 9 years 11 months, 18 were between 10 years and 15 years 11 months, and nine were over 16. The interviews which lasted from one to two and a half hours, were tape recorded with the parents' permission and were subsequently analysed.

Findings

Many parents had experienced long and distressing delays in obtaining an accurate diagnosis of Duchenne muscular dystrophy. Individual experiences differed; some parents became concerned when their sons were only 6 months old, while others did not become concerned until their sons were 4 or 5 years old. A small group of parents had not noticed anything wrong and were prompted to seek medical help by friends or teaching staff.

Typically the parents first sought advice from their general practitioners. Almost all were sent away either with a reassurance that the symptoms were "within the normal range of development" or with some rather caustic remarks about fussing unnecessarily. The time between the parents first becoming aware of symptoms that caused them anxiety and the final diagnosis averaged 2·5 years. Two patterns of experience after referral to a specialist were reported: either the diagnosis of muscular dystrophy and subsequent specification of the type of dystrophy was made relatively quickly or the boy was initially diagnosed as having something else, such as mental handicap, dislocated hips, or cerebral palsy. Parents who were originally given a mistaken diagnosis emphasised their difficulty in adjusting to the initial diagnosis only to be confronted later with a more devastating diagnosis.

A paediatrician eventually informed most parents of the diagnosis of Duchenne muscular dystrophy; but in seven of the 53 families the diagnosis was given by a paediatrician and social worker together, by an orthopaedic surgeon, or by a general practitioner. In one case the paediatrician had "about 10 students" with him. Only in 18 of the 53 families were both parents told together. Many of the parents who had been alone when told described how their distress was heightened by having to break the news to their husbands or wives.

Parents remember little of what they are told at the initial diagnosis,[1] and what they recalled was sketchy and at times inaccurate. Lack of follow up to the initial communication in some cases resulted in complaints by parents that they had not been given adequate information. Several of the parents recognised and commented on the difficult job that the paediatrician has in communicating such a diagnosis. In 15 of the 53 families, however, the parents were "not satisfied" with the way in which they had been told the diagnosis of Duchenne muscular dystrophy.

Analysis of the data showed that these parents had experienced an average delay of 3·46 years in obtaining an accurage diagnosis compared with only 2·04 years in the 36 "satisfied" parents ($p = 0{\cdot}05$; Student t test, two tailed test).

Discussion

Throughout the interviews a recurrent theme was that parents thought that they had been given less than adequate information by professionals. Undoubtedly in many of these cases the

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paediatrician had in fact given the parents a full, detailed, and accurate account of the disease and prognosis, but parents often cannot absorb the information given to them at this time. They may actively reject and deny what is being said to them. To help parents fully and completely understand the implications of their son's diagnosis a series of meetings between an informed professional and both parents seems to be necessary.

Some parents were lucky: "They told us straight away what it was, and the outcome. . . . They said 'Come up next week to talk about it.' Then he said, 'Go home and think about it and write out a long list of questions and come back and ask us them.' He then phoned our general practitioner who came over to talk to us."

Many, however, had a one off exchange with a paediatrician and little or no follow up.

One common policy of paediatricians was to tell the parents to contact them if they needed further help. Such "loose" offers of help do not take into account the fact that many parents may not feel able to seek out help in the early stages of acceptance. Of those parents who had been told that they could telephone and make an appointment for a further consultation, none took this action—despite there being things they would have liked to discuss.

Recommendations

Though there is clearly no single best way to tell the parents of a child that he has Duchenne muscular dystrophy the suggestions of parents who have been through this experience are summarised below.

(1) Most parents want to know if there is something wrong with their child as soon as possible. Evidence from parents of handicapped children shows that most have strong feelings that early telling is desirable.[2-5]

(2) Most parents prefer to be told the diagnosis together.

(3) Parents should be given some privacy when told the diagnosis. The number of people present should be kept to a minimum. Ideally the parents should be allowed to have some time on their own after the initial shock of the diagnosis to allow some release of emotions in private.

(4) The process of notification should aim to give parents full and balanced information and advice. Parents have different opinions about how much of the prognosis should be explained during the initial notification session, but most favour being "told straight" so that they were aware of all the implications from the start.

(5) As many parents will be unable to take in much of what they are told at the first notification of the diagnosis a series of contacts should be planned.[1]

(6) It may not be appropriate or necessary for the paediatrician to be the only source of support and information to the family immediately after the notification. Social workers and other professionals specialising in neuromuscular diseases who have a clear understanding of the disease and its implications could provide good follow up support to families who have recently learnt the diagnosis.

(7) At each meeting with newly notified parents it may help to suggest that there are sure to be many questions that the parents will want to ask and to encourage them to ask questions or voice their worries and uncertainties.[6]

(8) Between appointments with professionals it may help to encourage parents to write down every question/problem that comes to mind in the interim period. Parents sometimes forget their questions when face to face with doctors and other professionals.

(9) The offer of contact with another parent of a child with Duchenne muscular dystrophy may help. The parents had strong feelings both for and against this form of support.

I would like to acknowledge the help of all the parents who took part in this study and the continued support and guidance of Dr D Gardner-Medwin, consultant paediatric neurologist, Regional Neurological Centre, Newcastle General Hospital, Newcastle upon Tyne, and Dr G Hosking, consultant paediatric neurologist, Ryegate Centre, Tapton Crescent Road, Sheffield.

This study was supported by a grant from the Muscular Dystrophy Group of Great Britain, Nattress House, 35 Macaulay Road, London SW4 0QP.

References

1 National Association for Mental Health Working Party. The birth of an abnormal child: telling the parents. *Lancet* 1971;ii:1075-7.
2 Tizard J, Grad JC. *The mentally handicapped and their families*. London: Oxford University Press, 1961.
3 Drillien CM, Wilkinson EM. Mongolism: when should parents be told. *Br Med J* 1964;ii:1306.
4 Berg JM, Gilderdale S, Way J. On telling parents of a diagnosis of mongolism. *Br J Psychiatry* 1969;**115**:1195-6.
5 Carr J. Mongolism: telling the parents. *Develop Med Child Neurol* 1970; **12**:213-21.
6 Klauss NH, Kennell JM. Parent to infant attachment. In: *Recent advances in paediatrics*. Vol 5. London: Churchill, 1976.

(*Accepted 31 December 1982*)

Clinical curio: giant doses for giants?

Acromegalics are known to have large kidneys and an increased renal blood flow and glomerular filtration rate[1]; this could theoretically affect their handling of antibiotics. Recent successful treatment of such a patient with *Proteus mirabilis* meningitis after transsphenoidal hypophysectomy with gentamicin (and ceftazidime) has emphasised that conventional doses of gentamicin are often insufficient when renal function is good[2] and that there is no substitute for regular carefully controlled serum assays.[3,4]

The patient was a 51 year old man weighing 75 kg with a blood urea of 5·9 mmol/l (35·5 mg/100 ml) and a creatinine clearance of 111 ml/min. A loading dose of 160 mg gentamicin with maintenance doses of 120 mg eight hourly seemed a reasonable regimen which accorded with that predicted by the nomogram used in this hospital.[5] Trough and peak concentrations of < 1 and 4·5 mg/l respectively were achieved, and increasing the dose to 160 mg gentamicin eight hourly still produced trough concentrations of only < 1 mg/l and peaks of only 5·6 and 6·2 mg/l. A regimen of 200 mg gentamicin eight hourly was advised—the largest dose ever prescribed at St Thomas's Hospital in 16 years of gentamicin usage. This achieved therapeutic concentrations with troughs between 1 and 2·1 mg/l and peaks between 7·3 and 9·8 mg/l, which were monitored daily for one week. Intravenous ceftazidime was given in addition to gentamicin in a dose of 2 g six hourly. This regimen gave trough and peak levels of 9·9 mg/l and 78 mg/l respectively.

Twenty two other patients have been treated at St Thomas's with ceftazidime. Of those treated with 1 g eight hourly (the most frequently used regimen) nine had comparable renal function (with blood ureas of less than 7 mmol/l (42 mg/100 ml)) and the mean serum concentrations of ceftazidime in these patients were 9·6 mg/l (trough) and 58 mg/l (peak). This patient thus required a much larger dose of ceftazidime to achieve similar levels. These assay results serve more to emphasise the difficulties of predicting adequate dosage regimens in the individual than to suggest that all acromegalics require higher doses of antibiotics, and they underline the need for regular carefully controlled serum assays.—R P EVANS, senior house officer, and SUSANNAH EYKYN, consultant microbiologist, London.

1 Hall R, Anderson J, Smart GA, Besser M. *Fundamentals of clinical endocrinology*. 3rd ed. London: Pitman Medical, 1980.
2 Wise R, Reeves DS. Gentamicin dosage. *Br Med J* 1972;iv:732.
3 Kaye D, Levison ME, Labovitz ED. The unpredictability of serum concentrations of gentamicin: pharmacokinetics of gentamicin in patients with normal and abnormal renal function. *J Infect Dis* 1974;**130**:150-4.
4 Barza M, Brown RB, Shen D, Gibaldi M, Weinstein L. Predictability of blood levels of gentamicin in man. *J Infect Dis* 1975;**132**:165-74.
5 Mawer GE, Ahmad R, Dobbs SM, Tooth JA, Lucas SM, McGough JG. Prescribing aids for gentamicin. *Br J Clin Pharmacol* 1974;**1**:45.