Attitudes of mothers to neonatal screening for Duchenne muscular dystrophy

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Early diagnosis of Duchenne musclar dystrophy is crucial if genetic counselling is to be offered to women at risk of carrying the relevant gene. The age at diagnosis is still unacceptably late,[1] and we recently showed that screening programmes in infants are inadequate[2]; thus interest in neonatal screening has increased. Screening of infants has shown that assay of creatine kinase activity fulfils the criteria that should be satisfied by any technique used in neonatal screening programmes.[3] Despite the lack of effective treatment neonatal screening programmes exist in some countries[4] and the prevention of secondary cases has been put forward as a justifiable reason for introducing neonatal screening,[5] although there are ethical problems with this argument. We determined the attitudes of mothers towards neonatal screening for Duchenne muscular dystrophy.

Subjects, methods, and results

A structured questionnaire was administered by one interviewer to all mothers of newborn babies during one month in one obstetric unit. It was piloted initially on 20 subjects to remove any ambiguities in the questions. There were eight questions: the first three dealt with knowledge of existing neonatal screening programmes, the next three with Duchenne muscular dystrophy, and the final two with general attitudes towards early diagnosis of handicap and termination of pregnancy for medical reasons. A brief paragraph explaining the nature of Duchenne muscular dystrophy was included in the questionnaire so that the subjects would have some basic knowledge on which to make their decision.

Altogether 201 mothers (84% of the total delivered) were interviewed. The table gives their responses to the eight questions.

Responses to questionnaire on neonatal screening and its implications

	All mothers (n=201)	Multiparous women (n=109)	Primiparous women (n=92)
Knew of existing screening programme	137	94	43
Had heard of:			
Hypothyroidism	38	16	22
Phenylketonuria	36	24	12
Muscular dystrophy	147		
Duchenne muscular dystrophy	54		
Would accept screening test for Duchenne muscular dystrophy	189*	103	86
Would want to know at birth if their child was handicapped:			
Definitely	151	77	74
Probably	28	20	8
Not sure	14	7	7
Probably not	4	1	3
Definitely not	4	4	
Would consider termination of pregnancy for medical reasons:			
Definitely	58	33	25
Probably	84	46	38
Not sure	38	19	19
Probably not	6	3	3
Definitely not	14	9	5

*Mothers of 90 boys (93%) and 99 girls (95%).

Comment

We found that only 68% of the mothers were aware that infants were screened neonatally. Not surprisingly, many more multiparous than primiparous mothers were aware of such screening. Knowledge of the conditions screened for was poor, even among multiparous mothers. Many more women had heard of muscular dystrophy than of hypothyroidism and phenylketonuria, possibly owing to the widespread fund raising activities of the Muscular Dystrophy Group.

By far the majority of mothers said that they would opt for screening for Duchenne muscular dystrophy. Although only boys would be screened, there was no difference in responses between mothers of boys and girls. As the questionnaire was administered we were able to ensure that the subjects clearly understood that the condition was untreatable and that the main reason for having the screening test was to prevent the birth of further affected babies in the family.

The answers to the general questions showed that most women would want to know soon after birth whether their baby had a handicapping disorder. As the puerperium is a sensitive period in which to question attitudes to termination of pregnancy there is some doubt about the validity of this type of question and the reliability of responses. This may have some bearing on the proportion of mothers who would be prepared to accept termination. Nevertheless, most of the women said that they would consider termination of pregnancy for medical reasons.

These findings have implications for anyone considering establishing a neonatal screening for Duchenne muscular dystrophy. As a few women would not want their child to be screened they would have to be allowed to opt out of such a screening programme. The mothers' poor awareness of existing neonatal screening suggests that little effort has been made to inform them about it. Considerable effort will be needed to enhance awareness and understanding in a society increasingly aware of and sensitive to the importance of full information.

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