Editorial

Psychosocial effects of screening for somatic risk: the Swedish α_1 antitrypsin experience

During 1972-74 200 000 Swedish newborn children were screened for α_1 antitrypsin deficiency in association with routine screening for phenylketonuria.¹ The parents of infants with the *PiZZ* or *SZ* forms of α_1 antitrypsin deficiency were informed of the child's deficiency by telephone or letter when the infants were 2-4 months of age. The family then had an appointment with a special doctor to discuss the α_1 antitrypsin deficiency, and most families made subsequent visits until the child was 4 years of age, the visits including measurement of liver enzymes. The main purpose of this early identification and follow up was to study the natural history of α_1 antitrypsin deficiency in childhood and to provide an opportunity to protect the affected children from concentrated air pollutants (mainly cigarette smoke), in the hope of preventing or postponing lung disease in adulthood.

Alpha₁ antitrypsin deficiency has special characteristics that may have implications for parents' reactions to its identification in their newborn children. At the time we did our study α_1 antitrypsin deficiency was a fairly new and generally unknown condition (for many medical personnel and most parents); in addition, very few children with α_1 antitrypsin deficiency are ill from the condition and no externally observable effects of the deficiency will appear in most individuals affected until adulthood. In view of the uncertainty about the nature and implications of α_1 antitrypsin deficiency a range of psychological reactions might therefore be expected to occur among parents.

As parents were being informed and the family followed up during the study, some paediatricians reported that identification of the child's α_1 antitrypsin deficiency appeared to have had considerable adverse psychological consequences for the parents and for the parent-child relationship. Neonatal screening for α_1 antitrypsin deficiency was discontinued in 1974, and a systematic investigation of the psychological and psychosocial consequences of the screening programme was initiated when the children were about 5 years of age. A representative sample of families of 61

clinically healthy *PiZZ* children were compared with 61 families of control children without α_1 antitrypsin deficiency who were carefully matched demographically and with 183 control families largely selected at random for demographic comparisons.² The α_1 antitrypsin deficiency and matched control families were studied in their homes by interviews with parents and observations of mother-infant interaction, and all three groups were studied by medical and population register records.

Most parents of the children with α_1 deficiency were found to have reacted negatively (usually with fear and anxiety) to the first news of the child α_1 deficiency. These reactions were typically strong and long lasting.³ Most parents thought that the first information about α_1 antitrypsin deficiency was unclear and inadequate, and perceived the deficiency as representing an immediate, serious threat to the child's health. About one third of the parents felt relief after the first appointment with the specialist doctor, at which they received additional information about α_1 antitrypsin deficiency.⁴

At follow up five to seven years later parental reports suggested that there were no negative long term consequences for the parents of children with α_1 antitrypsin deficiency with respect to reproduction rates,⁵ marital status and social class level,⁵ view of their life situation,⁶ attitude to themselves as parents,⁷ view of the child's personality and behaviour,⁸ attitudes and feelings towards the child,⁹ relationship with the child in everyday life,¹⁰ concern for the child's current health, or attitudes towards the paediatric health services in general.¹¹ When compared with matched control families, however, both mothers and fathers of the children with α_1 antitrypsin deficiency viewed their affected children as more sickly than other children¹¹ (not confirmed by systematic study of their medical records¹²). Mothers reported significantly poorer mental and physical health during the year before follow up, apparently as a consequence of stress associated with the finding of α_1 antitrypsin deficiency in their child.⁶ In addition, the children showed a notable increase in problematic behaviour (for example, aggression, inhibition, tension) when interacting

with their mothers in the context of a structured test.¹³

At follow up, about half the mothers and one third of the fathers were judged by a psychiatrist to have made a poor or very poor emotional adjustment to the child's α_1 antitrypsin deficiency. This was reflected in problematic negative feelings toward the child's deficiency and adverse consequences for the parents' relationships with other family members.¹⁴ Parents' long term emotional adjustment was unrelated to their demographic, familial, or personal characteristics at the time the child's α_1 antitrypsin deficiency was identified; poor adjustment was significantly predicted in both mothers and fathers by the development of a psychic crisis reaction when the child's deficiency was identified, and in mothers by repeated medical contacts regarding α_1 antitrypsin deficiency that were experienced as being negative (T Rosenlund *et al*, unpublished reports). A "positive" contact with a physician at the first appointment for discussing α_1 antitrypsin deficiency was an important determinant of the mother's good long term emotional adjustment; fathers adjusted better if they "denied" the finding of α_1 antitrypsin deficiency and discredited the medical system's competence and credibility regarding it.

Most parents said that they welcomed the fact that the child's α_1 antitrypsin deficiency had been identified at such an early age.⁴ Early identification did not, however, have a desirable effect on the one relevant risk behaviour factor, parental smoking. At follow up one or both parents smoked in more than half of the homes concerned; mothers of children with the deficiency smoked as frequently as control mothers, whereas fathers of these children smoked twice as frequently as control fathers (unpublished data).

Thus a cost-benefit analysis of this neonatal screening for α_1 antitrypsin deficiency showed notable psychological and psychosocial "costs" and an absence of "benefits" with regard to parental smoking during the child's early life.¹⁵ The extent to which an improvement of medical contact procedures could minimise the costs and maximise the benefits of identification of such a somatic risk condition is unknown. Further improvements in medical contact procedures and the provision of adequate psychological support for parents will be necessary if screening is to be justified by clinical benefits after early identification of risk conditions.

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References

- 1 Sveger T. Liver disease in alpha₁-antitrypsin deficiency detected by screening of 200 000 infants. *N Engl J Med* 1976;294:1316-21.
- 2 McNeil TF, Thelin T, Aspegren-Jansson E, Sveger T, Harty B. Psychological factors in cost-benefit analysis of somatic prevention. *Acta Paediatr Scand* 1985;74:427-32.
- 3 Thelin T, McNeil TF, Aspegren-Jansson E, Sveger T. Psychological consequences of neonatal screening for α_1 -antitrypsin deficiency. Parental reactions to the first news of their infants' deficiency. *Acta Paediatr Scand* 1985;74:787-93.
- 4 Thelin T, McNeil TF, Aspegren-Jansson E, Sveger T. Psychological consequences of neonatal screening for α_1 -antitrypsin deficiency (ATD). Parental attitudes toward "ATD-check-ups" and parental recommendations regarding future screening. *Acta Paediatr Scand* 1985;74:841-7.
- 5 Thelin T, McNeil TF, Sveger T. Identifying children at high somatic risk (α_1 -antitrypsin deficiency): possible long-term effects on parents' reproduction, marital status and social class level. *Acta Psychiatr Scand* 1985;71:636-43.
- 6 Thelin T, McNeil TF, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible long-term effects on the parents' views of their own health and current life situation. *Acta Psychiatr Scand* 1985;71:644-53.
- 7 Thelin T, McNeil TF, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible long-term effects on the parents' attitudes toward themselves as parents and toward having more children. *Acta Psychiatr Scand* 1985;72:74-80.
- 8 McNeil TF, Thelin T, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible long-term effects on parental conceptualizations of the children's personality and behaviour. *Acta Psychiatr Scand* 1985;72:498-504.
- 9 McNeil TF, Thelin T, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible long-term effects on the parents' attitudes and feelings associated with the child. *Acta Psychiatr Scand* 1986;74:341-6.
- 10 McNeil TF, Thelin T, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible long-term effects on the parents' relationship to the child. *Acta Psychiatr Scand* 1986;74:347-52.
- 11 McNeil TF, Thelin T, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: possible effects on the parents' views of the child's health and parents' relationship to the pediatric health services. *Acta Psychiatr Scand* 1985;72:491-7.
- 12 Sveger T, Thelin T, McNeil TF. A risk group's visits to medical services: five years' usage of well-baby and medical clinics by a group of children with α_1 -antitrypsin deficiency. *Acta Psychiatr Scand* 1985;74:782-6.

- 13 McNeil TF, Harty B, Thelin T, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: long-term effects on mother-child interaction. *Acta Psychiatr Scand* 1986;**74**:555–62.
- 14 Thelin T, McNeil TF, Aspegren-Jansson E, Sveger T. Identifying children at high somatic risk: parents' long-term emotional adjustment to their children's alpha₁-antitrypsin deficiency. *Acta Psychiatr Scand* 1985;**72**:323–30.
- 15 Thelin T. *Psychological effects of neonatal screening. A study of parents of children at high risk for serious illness in adulthood due to alpha₁-antitrypsin deficiency*. Doctoral dissertation, University of Lund, 1985.