Heterozygous familial hypercholesterolaemia (FH) is a common autosomal dominant inherited metabolic disease with a prevalence of 1 in 500 in most Western countries. People with FH experience an increased risk for coronary artery disease (CAD) and excess mortality especially at a young age. Until recently the diagnosis of FH was based on clinical signs and symptoms alone. These included increased cholesterol concentrations, in particular of LDL-cholesterol, in combination with the presence of tendon xanthoma, corneal arcus, xanthelasmata and a history of early CAD. Frequently FH was diagnosed after a first cardiac event.

GENETIC SCREENING

The discovery of LDL receptor gene mutations in clinically diagnosed familial hypercholesterolaemia (FH) patients and the consequent development of DNA tests for these mutations, enabled the diagnosis of FH in pre-symptomatic patients. In principle, the availability of effective treatment for FH made screening of relatives of identified FH patients an attractive strategy to reduce the risk of coronary artery disease (CAD).

In 1994, such a family-based genetic screening programme for FH was implemented in the Netherlands. This programme is targeted on at risk relatives (over 16 years of age) of clinically diagnosed FH patients with a known mutation in the LDL-receptor gene, also called index patients. First degree relatives of index patients are offered genetic testing for the mutation found in the index patient. If these relatives carry the mutation, their first degree relatives are screened. Mutation carriers are referred to their general practitioner with the advice to have a further specialist examination at a lipid clinic.

SOCIAL CONSEQUENCES OF GENETIC SCREENING

Participating in such a genetic screening programme may induce other effects than the aimed health benefit because of the reduction of CAD risk. As a result of screening there may be negative social consequences, such as problems with regard to the access to insurance. In the Netherlands, as in most other countries, health insurance is compulsory. The health system in the Netherlands can be classified as a Sickness Insurance model. The Netherlands has developed a mixed health care system in which approximately 70% of the population is covered for their medical costs (except chronic residential care) through an income related social health insurance, and the remaining 30% through private health insurance. Insurance companies do not always accept people who apply for insurance, but a special insurance arrangement is available for persons difficult to insure. If, however, a person has to leave the social health insurance plan as a result of a higher income, national rules enforce acceptance by a private insurer. In the past decade there has been a transition from many sick funds and private health insurers into a few large financial institutions. These companies offer a wide array of work disability and life insurance packages, often in combination with mortgages. People who apply for insurance have to fill out a health questionnaire. Coverage of claims depends on truthful response to this assessment of prior risks. However, restrictions exist with regard to enquiries about genetic test information. In the Netherlands, a moratorium has been declared on the use of genetic test results. In this moratorium, insurance companies state that below an amount of 300 000 Dutch guilders (approximately £85 000) for life insurance and below an amount of 60 000 Dutch guilders for the first year and 40 000 Dutch guilders for the second and following years in work disability insurance, the results of genetic tests do not have to be supplied (including information with regard to the suffering or death of family members because of serious untreatable familial disorders). The Act on Medical Examinations, which came into force in 1998, contains the same conditions.

Many European countries have no legislation or guidelines on insurance and medical examinations with respect to genetic testing. Other countries (for example, France) have guidelines and have imposed a moratorium on the use of genetic tests. In the UK, guidelines on genetic testing are proposed by the insurance industry, rather than by the government.

The issue of this paper is whether such a moratorium on the use of genetic tests or legislation...
how did problems relate to the medical condition: were
• which problems were the most prevalent, if any;
• addressed:
Examinations were into force. The following questions were
a period in which the moratorium and/or the Act on Medical
experiencing problems in the access to insurance.

Table 1 Problems with insurance related to clinical
risk: mutation and cholesterol level

<table>
<thead>
<tr>
<th>Clinical risk</th>
<th>Problems</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mutation</td>
<td>Cholesterol level</td>
</tr>
<tr>
<td>No</td>
<td></td>
<td>≤6.5 mmol/l</td>
</tr>
<tr>
<td></td>
<td></td>
<td>&gt;6.5 mmol/l</td>
</tr>
<tr>
<td>Yes</td>
<td></td>
<td>≤6.5 mmol/l</td>
</tr>
<tr>
<td></td>
<td></td>
<td>&gt;6.5 mmol/l</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

protects participants of a genetic screening programme from
experiencing problems in the access to insurance.

EMPIRICAL STUDY
We conducted an empirical study among people screened for
FH in the Netherlands by the programme mentioned above, in
a period in which the moratorium and/or the Act on Medical
Examinations were into force. The following questions were
addressed:
• to which extent do people screened for FH, run into
problems when applying for insurance (depending on the
test result);
• which problems were the most prevalent, if any;
• how did problems relate to the medical condition: were
problems attributable to a genetic mutation for FH, to an
increased cholesterol concentration, or both. We looked at
an increased cholesterol concentration as this will be the
most likely confounder in this population of mainly asympto-
matom carriers and non-carriers.

From approximately 1000 persons screened between 1
January 1994 and 31 December 1997, 350 people aged 20 to 60
years with known test results were selected from the
registration. We aimed for an equal distribution of age and
gender in the group with (n=175) and without (n=175) a
mutation for FH. The age range was chosen for two reasons.
Life insurance and mortgages are predominantly relevant for
this age range, and cholesterol levels for those aged over 60
often exceed cut off points used by insurance companies.

Administrative data were made available by the organisa-
tion responsible for the screening programme (the StOEH
foundation). To protect privacy, the StOEH, rather than the
investigators, invited selected persons to participate by means
of a postal questionnaire. Questionnaires could be returned to
the StOEH. One reminder was sent two weeks after the first
questionnaire. The study was approved by the medical ethical
committee of the Academic Medical Centre.

Of the 350 addresses, 327 apparently were valid. Of the 327
people who received the questionnaire, 202 returned the
questionnaire (61% response). More women than men
returned the questionnaire (116 versus 86). Of the 202
respondents, 46 people applied for insurance in the period
between being screened and the study (23%). Of those 46
people who tried to get insurance, 17 encountered problems
(37%; see table 1).

The type of problem most often encountered was the
requirement to pay a higher premium, the requirement to
undergo additional medical tests, the requirement to let the
insurance company scrutinise medical records, and complete
rejection of the application for insurance. If stated, the
argument of the insurance companies to support their policy
was the fact that the applicant had FH. Surprisingly, most (13
of 17) people who encountered problems applied for a life

insurance well below the cut off points stated in the morato-
rion and the Act on Medical Examinations, which implies
that the applicants were not obliged to provide the insurance
company with information on genetic testing.

The question is whether the encountered problems occur
because these people have a genetic mutation for FH. Another
possibility is that problems occur because of their increased
cholesterol level, as this is the most likely confounder in this
population of mainly asymptomatic carriers and non-carriers.
The table shows the association between an increased choles-
terol level, the presence of a genetic mutation for FH and the
occurrence of problems among the 46 respondents who

We apply a 6.5 mmol/l cut off in cholesterol concentration
because insurance companies consider people with a choles-
terol level above this cut off point to be at increased risk of
death. We used log linear analysis with having problems as the
dependent variable and the mutation and cholesterol concen-
tration as independents. Correlation between the independent
variables was assumed. We looked for the most economic
model that would fit the data—that is, the model with the
fewest interactions. The model in which only the mutation is
associated with having problems fitted the data (likelihood
ratio χ²=2.32, df=2, p=0.31), while the model in which only the
cholesterol level is associated with having problems did not
(likelihood ratio χ²=6.70, df=2, p=0.035). Therefore, from
this analysis it seems that encountered problems primarily
relate to the presence of the FH mutation. If this were true,
this policy would be at discrepancy with the current scientific
state of the art, which acknowledges the cholesterol (level) to
be the transmitter or carrier of the higher mortality risk asso-
ated with FH. However, given the small sample size and
therefore the limited power of this analysis, as well as the lack
of data on other possible confounders, a final conclusion
regarding this issue cannot be drawn.

DISCUSSION
This empirical study has shown that in the Netherlands,
which combines relevant guidelines and legal arrangements
on the use of genetic testing in insurance affairs, participants
of a genetic screening programme encounter unanticipated
insurance problems. Although the number of people exposed
to an insurance procedure is small (46) because of the limited
follow up time of tested persons, the proportion of those
encountering problems (a third) is worrisome, even if one
accounts for some selective response. Moreover, others
reported similar data.2

Even more disturbing was the presence of problems in cases
where the Act on Medical Examinations explicitly rules out a
role for genetic test information. From our study it is not clear
why these problems occur—that is, whether insurance
companies ask questions regarding genetic testing or ques-
tions that can be read as such, or that individuals
themselves give more information than asked for.

We conclude that guidelines and legislation on genetic
information are but a prerequisite and that education of all
involved is equally important.

Key points
- In the Netherlands guidelines and legislation exist on the
  use of genetic test information.
- This study shows that participants of a genetic screening
  programme still encounter unanticipated insurance prob-
  lems.
- It is not clear why these problems occur: whether insurance
  companies ask questions regarding genetic testing or ques-
  tions that can be read as such, or that individuals
  themselves give more information than asked for.
- We conclude that guidelines and legislation on genetic
  information are but a prerequisite and that education of all
  involved is equally important.
for the relatively uneducated. When asked for, only 14% of the individuals knew about the Act on Medical Examinations or the moratorium. The line of questioning or the repeated statement in questionnaires that insurance coverage is lost if the information supplied is incorrect may induce over-response by the applicant. Furthermore, compared with questionnaires designed for scientific purposes (for example, health risk measurement in surveys), the existing insurance questionnaires frequently appear ambiguous, providing the applicant no clues as how to interpret questions.

Our main conclusion is that implementation of the Act on Medical Examinations requires more control on its procedural execution. Moreover, the participants of a genetic screening programme, the public in general, but also those engaged in the insurance acceptance process should be educated about the existing guidelines regarding genetic testing. At this stage responsibilities are ill defined. Recently, the screening organisation included explicit information on the issue before the testing procedure. Our case study shows that the presence of national guidelines and regulations on the (mis)use of genetic information is but a prerequisite for appropriate use of genetic information in the context of insurance. Education of all involved, including the screenee/patient is equally important.

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Conflicts of interest: none.

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Authors’ affiliations
P J Marang-van de Mheen, M C van Maarle, M E A Stouthard, Department of Social Medicine, Academic Medical Centre, Amsterdam, the Netherlands

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Getting insurance after genetic screening on familial hypercholesterolaemia; the need to educate both insurers and the public to increase adherence to national guidelines in the Netherlands

P J Marang-van de Mheen, M C van Maarle and M E A Stouthard

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