Familial hypercholesterolemia screening in the Netherlands: psychological and social consequences
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Chapter 5

Risk Perception of Participants in a Family Based Genetic Screening Program on Familial Hypercholesterolemia

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Abstract

The aim of this article is threefold. First, we describe the accuracy people’s risk perception who have been screened on Familial Hypercholesterolemia (FH) in a family-based screening program. Second, we identify factors that modify risk perception. Finally, we show the influence of risk perception on subsequent preventive behavior.

The risk perception of 556 (677 participants, overall response = 82%) screenees was measured by postal questionnaires on 3 occasions: at screening, 3 days and 7 months after the test result was reported to the patient. Presentation of the risk was pre-categorized and given both as numerical (1 in x) and as verbal probability. In addition, medication use and attitudes towards gene therapy were determined 7 months after screening.

On average, the screenees underestimated their numeric risk of having FH and getting a myocardial infarction (MI). Furthermore, FH-positive screenees perceived they were at greater risk of MI than FH-negatives, and screenees with the highest actual risk used medication more, perceived a greater risk and opted more often for future gene therapy.

Risk perception of having FH was influenced by cholesterol level, while MI risk perception was affected by age, education, cholesterol level, and cardiovascular disease (CVD) in the family. We conclude that FH-positive screenees correctly perceive a higher risk of getting a heart attack than do FH-negative screenees. Screenees did not believe that MI was inevitable and risk perception was associated with both medication use and the intention to opt for gene therapy, but not with other preventive measures. Thus, genetic risk notification seems to be acceptable and does not lead to aversion to preventive behavior.
Introduction

Genetic information is currently used for risk estimation of common diseases and this use will probably increase in the near future.[Bell, 1998] Consequently, genetic family and population screening has gradually become part of regular patient care. The main goal of genetic screening is to make people aware of their genetic risk, which in turn should stimulate specific preventive health behaviors.[Breslow et al., 1997; Womeodu and Bailey, 1996] However, while genetic risk notification should raise risk awareness, it should not distort the perception of actual and future risk.[Marteau, 1999] Despite extensive risk perception research (see e.g. Tversky and Kahneman [1974] and Van der Pligt [1998]), it is not clear whether providing genetic risk information will motivate people to engage in preventive health behavior or will even have the opposite effect. It could motivate to change behavior by strengthening the belief that current behavior combined with the genetic risk is putting oneself at increased risk of the disease; additionally, it could strengthen the belief that treatment will be more effective when it is recommended on basis of genetic information.[Marteau and Lerman, 2001] However, it has been observed that people who have a (non-genetic) family history of heart disease are not more or less likely to engage in risk reducing behavior.[Becker and Levine, 1987] Moreover, other studies showed that heritability and genetic risks could be viewed as being uncontrollable and inevitably leading to disease, thus making treatment useless.[Troein et al., 1997; Senior et al., 1999] Overall, there is currently no evidence that providing genetic risk information increases people's motivation to change health behavior more than would be achieved with non-genetic information and for some people, genetic information may even reduce the motivation to change behavior.[Marteau and Lerman, 2001]

Familial hypercholesterolemia (FH) is a genetic risk factor for coronary artery disease (CAD), with an estimated frequency of 1 in 500 persons (in Western countries).[Goldstein et al., 1973; Goldstein and Brown, 2001; Motulsky, 1989] It results from a mutation that affects the function of the receptor that normally removes low-density-lipoprotein from the plasma, and thus can result in an accumulation of plasma-cholesterol and consequently in an excess CAD mortality.[Scientific Steering Committee 1991; Goldstein et al., 1973; Goldstein and Brown, 2001; Motulsky, 1989] Currently, over 700 different mutations are identified.[Anonymous, 2002] When either the plasma total cholesterol or LDL cholesterol level is used as a genetic marker, the FH gene is highly penetrant at all ages.[Goldstein et al., 2001] However, some recent studies show different LDL cholesterol levels for different mutations in the LDL gene.[Bertolini et al. 2000; Chaves et al. 2001; Slimane et al. 2001; Jansen et al. 2002] In FH a delay between –asymptomatic– hypercholesterolemia and overt CAD exists. Preventive measures are effective in this asymptomatic stage.[Anonymous, 1994 and 1998; Scientific Steering Committee, 1999; Downs et al., 1998; Hoogerbrugge , 1998; Shepherd et al., 1995] The availability of DNA diagnosis at an asymptomatic stage and effective lipid-lowering therapy make genetic screening on FH feasible.

In 1994, an FH-screening program started in the Netherlands in a provisional setting with a parallel, independent evaluation study. The evaluation study comprised, among other things, an evaluation of risk perception of screenees in terms of the perceived probability of getting a heart attack later in life, depending on risk status (FH-positive or negative) and personal characteristics. Furthermore, to determine whether risk perception was associated with preventive behavior.
The evaluation focussed on the perception and representation of information by FH-screenees, the change of risk perception over time, as influenced by test result and other information/experience, and the consequential influence of risk perception on actual risk reduction (cholesterol-lowering medication use, stated attitude towards gene therapy, smoking cessation and losing weight). The main aims of our study were to establish whether: 1) disease experience, e.g. cholesterol-lowering therapy and deceased family members would increase risk perception in general; 2) screenees would incorrectly ignore the risk difference between having FH and getting FH-related CAD; and 3) increased awareness of risk would lead to more risk reducing measures.

By carefully measuring risk perceptions in a large, relevant cohort, we hoped to contribute to quality-improvement of family-based screening programs.

**Material and Methods**

**Screening program**
The Dutch screening program actively approaches first- and second-degree relatives of index patients (i.e. clinically diagnosed FH patients with a known mutation). The 'Foundation for tracing hereditary hypercholesterolemia' (Dutch acronym: StOEH) is responsible for this pedigree investigation ('cascade screening').[Umans-EckenhAUSEN et al. 1999; Umans-EckenhAUSEN et al. 2001] If relatives test positive, their first- and second-degree relatives are approached and offered testing, etc. Relatives are not tested for all known mutations, but only for the mutation found in the index-patient. Cholesterol is not measured within this screening program. All test results are communicated to the screenee by mail. Screen-positives get two additional letters: one directed to him/herself with the advice to consult the general practitioner (GP), and one to the GP, inviting the GP to refer the patient to a lipid clinic. This procedure requires the screen-positives to take the initiative to seek medical follow-up.

**Written information supplied in the screening program**
Before screening, relatives of FH-patients are approached by mail. This letter also comprises a leaflet, which gives more information about FH. As no exact figures are known about the penetrance estimates and consequent CHD risk, only a general description of FH is given. The leaflet explains that FH causes hypercholesterolemia, subsequently causes damage of the blood vessels, atherosclerosis, and eventually myocardial infarction (MI). Also, information about the chances of inheriting the gene defect is given: somebody with FH has a 50% chance to pass the gene defect to his or her child. Additional to this leaflet a specific booklet is included with the test result in the case the screenee tests positive. This booklet provides a detailed description of the biochemical mechanism of FH, information about inheritance and the risk of MI. It is stated that FH-positive screenees have a high risk to get an MI. Furthermore, the leaflet reassures the reader that hypercholesterolemia is treatable and that most FH-patients can normalize cholesterol levels with medication and diet, thus lowering the chance of getting an MI.

**Subjects**
Inclusion of the subjects in the study was between March and September 1998. The inclusion criteria were: informed consent to genetic testing and to our survey, and age 18 years or over. With consent of the participants, their FH-status was disclosed to the researchers. The study was approved by the medical ethical board of the hospital.
**Data collection**

For the study reported here, data were collected by means of three self-administered questionnaires (T0: at screening, before knowing the test result; T1 and T2: 3 days and 7 months after the test result, respectively). Risk perceptions were evaluated in all 3 questionnaires. The first questionnaire covered socio-demographic data, last year's prevalence of cardiovascular disease (CVD) manifestations and familial prevalence of CVD (a/o CVD death before the age of 50). In the third questionnaire body weight, height and smoking status were recorded, for both at the time of screening and 7 months after screening. Cholesterol level was asked in the first and third questionnaire. In the first questionnaire it was only asked whether people had hypercholesterolemia (yes, no, or I don't know), in the third questionnaire cholesterol level was asked in words (high, normal, low), and in numbers (mmol/l). Cholesterol-lowering medication use was asked in the first and third questionnaire and attitude towards gene therapy, should this become available, in the third.

Three prior risk strata were defined. High risk was defined by clinical hypercholesterolemia and/or cholesterol-lowering medication use; intermediate risk by an unknown cholesterol level and no cholesterol-lowering medication; and low risk by a normal cholesterol level while no cholesterol-lowering medication.

The risk perception of the screeners was studied, distinguishing between the probability of having FH and that of getting a heart attack later in life. The presentation of risk perception was pre-categorized and given both as numerical (1 in x) and verbal probability (see figure 2). Perceived risk of getting a heart attack later in life was subdivided into the perceived risk with and without treatment. Risk perception was studied before and after knowing one's FH status.

**Data Analysis**

Descriptive analyses were conducted and differences of the tested variables between groups were assessed by Pearson's Chi-square statistics and Student's t-test. For within-subject changes over time we used the Friedman test for categorical data. The relation at the time of screening between the set of clinical and socio-demographic independents on the one hand, and risk perception on the other was established. Risk perception was defined here as the perceived chance of having FH (at T0, hence before knowing the test result), and the perceived probability of getting a heart attack later in life when having FH and not being treated (both in numerical and verbal probability). Selected variables from the univariate analysis (p<0.1) were further offered to a standard multivariate multinominal logistic model (multinominal rather than standard logistic model as the dependents were polytomous). The Nagelkerke's R2 of the used models showed the explained variance.

Focussing only on FH-positive screeners, the association between medication use 7 months after screening and socio-demographic, clinical and risk perception variables was established. The variables were offered to a logistic regression model, using backward elimination method (pin=0.05; pout=0.1). Furthermore, the association between considering gene therapy and the above-mentioned set of variables was tested in a similar way. The analyses are based on N=647 when only variables from the first questionnaires are used (basic characteristics and screeners' expected FH-status), and based on N=556 in all other analyses. This statistical evaluation was performed using the SPSS version 10.0.07 for Windows.
Results

Response
In the evaluation study period, 720 people of the cohort met the inclusion criteria for our survey and were asked to participate (see Figure 1). Of those, 43 people did participate in the screening program but decided not to participate in our survey. This leaves 677 participants in the survey, of whom 647 sent back the first questionnaire and 556 (82%) sent back all three questionnaires. The risk perception questions were filled out nearly complete, with only 2-4% missings or uninterpretable responses per question.

Figure 1 Participation in survey

Lost to follow-up
There was no significant difference in age, sex, marital status, FH-status and educational level between the screenees lost to follow up (N=91) and the screenees who returned all three questionnaires (N=556). Of the 30 people who agreed to participate but did not send the first questionnaire back, no further information was available; these screenees were considered as non-respondents rather than as lost to follow-up.

Basic characteristics of the screenees
Table 1 presents the basic characteristics of the screenees. Overall, 46% were men and the mean age was 47 years. Furthermore, 56% had not previously heard of FH either in general or as occurring in their family, still 45% of these screenees reported first-degree family members with CVD, and 15% reported family members (total family) who died of premature CVD. Of all screenees, 3% reported to have CVD, 36% reported being hypercholesterolemic, 26% reported a normal cholesterol level, and the remaining screenees did not know their cholesterol level. After testing, 32% of our study population proved to be FH-positive.
Table 1 Basic Characteristics of the screenees (N=647)

<table>
<thead>
<tr>
<th></th>
<th>%</th>
<th>Range</th>
</tr>
</thead>
<tbody>
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<tr>
<td>Women</td>
<td>54</td>
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<tr>
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<td>(18-87 year)</td>
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<tr>
<td>Men (range)</td>
<td>48 year</td>
<td>(18-79 year)</td>
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<tr>
<td>Women (range)</td>
<td>47 year</td>
<td>(18-87 year)</td>
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<td>before screening (% yes)</td>
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<tr>
<td>(before screening)</td>
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<tr>
<td>through:</td>
<td></td>
<td></td>
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<tr>
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<td>63</td>
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<td>Stroke</td>
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<td>Diabetes</td>
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</tr>
<tr>
<td><strong>Premature CVD² deaths in family (% yes)</strong></td>
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<td></td>
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<td>FH positive</td>
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<td></td>
</tr>
<tr>
<td>FH negative</td>
<td>68</td>
<td></td>
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</tbody>
</table>

¹ Missings excluded in percentages
² In the first questionnaire the participants was asked whether they had a high cholesterol level. This level was not further specified.
³ Now or in the last 12 months
⁴ CVD death at the age of 50 or younger
Risk perception at screening and change over time

**Screenees’ expected FH-status**

At the time of screening (T0; N=647) almost 50% of the screenees rated their verbal chance of having FH as high or very high and 44% rated their numerical chance of having FH 1 in 3 and higher. In retrospect the FH-positive screenees appeared to have rated their chance of having FH significantly higher than the FH-negative screenees (p<0.001). This remained true if we excluded screenees with prior knowledge of their cholesterol level. Screenees in the highest prior risk stratum perceived a significantly higher risk than screenees in an intermediate or low prior risk stratum (p<0.001; figure 2).

**Figure 2 Risk perception of having FH and actual risk**

**Risk of getting a heart attack later in life**

In figure 3 the verbal and the numerical risk perception for getting a heart attack are shown (N=556). At T0 both the expected risk in case of a being FH-positive and being FH-negative were asked; from T1, the risk perception refers to one's actual FH-status. Both in verbal and numerical risk terms, and at all three points in time, the FH-positive screenees perceived a significantly higher risk of getting a heart attack later in life than FH-negative screenees (p < 0.05). Generally, this difference was also observed in the situation with treatment. The risk perception of the FH-positive screenees was significantly lower in the condition with treatment than without treatment, at both T1 and T2 (p<0.001). On average, both FH-positive and FH-negative screenees perceived a decreasing risk (verbal and numerical) over time (p<.05), although in verbal risk perception 16% of the FH-negative and 19% of the FH-positive screenees perceived a higher risk at T2 than at T0. In numerical risk perception these figures were 16% and 13%, respectively.
Factors associated with risk perception

**Screenees’ expected FH-status**

In verbal risk perception the younger screenees rated their chance of having FH higher than older screenees. Furthermore, screenees with hypercholesterolemia, higher education and/or CVD in the family (having first-degree family members with CVD and premature CVD deaths in the family) rated their verbal chance of having FH higher than screenees without these characteristics (N=556; all: p<0.05, except having first-degree family members with CVD: p = 0.08). These factors explained 41% of the variance. Results in numerical risk perception were much alike: with the exception of having heard of FH (added) and educational level (removed), the explanatory variables were the same, as was the explained variance.

**Risk perception of getting a heart attack later in life**

In verbal risk perception the older screenees rated their risk lower than the younger screenees. Furthermore, screenees with hypercholesterolemia, higher education and/or first-degree relatives with CVD perceived a higher verbal risk than screenees without those characteristics. Screenees who did not know their cholesterol level perceived the lowest verbal risk (N=556; p<0.05; explained variance: 22%). In numerical risk perception the associated variables were the same, except for age, but the explained variance was only 14%.
Association of risk perception and preventive behavior

Medication utilization
The medication utilization in the FH-positives rose from 58% at T0 to 77% T2 (p<0.001). In the FH-positive screenees, age, sex, cholesterol level in words and numbers, and numerical risk perception of getting a heart attack later in life with or without treatment were associated with medication utilization at T2 (p<0.05; explained variance: 56%). Screenees using cholesterol-lowering medication were significantly older, more often male, perceived a higher risk of getting a heart attack later in life (with and without treatment), had a higher cholesterol level and more often family members with CVD than screenees without these characteristics. Screenees with a substantial difference in risk perception between the situation with and without treatment (three or more categories difference and a lower risk perception in the situation with medication) are significantly more often using cholesterol-lowering medication (p<0.05).

Considering gene therapy
Of all FH-positive screenees, 69% would consider gene therapy when this would become available. Men, screenees who were living together or married, the higher educated, screenees who were not religious, screenees perceiving a higher risk of getting a heart attack later in life (with and without treatment) and screenees with hypercholesterolemia were more inclined to opt for gene therapy (p<0.05; explained variance: 43%).

Other preventive behavior
There was no significant change in smoking status and BMI, both in FH-positive and FH-negative screenees. At the time of screening 28% of the FH-positive and 26% of the FH-negative screenees smoked, compared to 23% and 26% at T2, respectively. The mean BMI at T0 was 24 and 25 kg/m2 in FH-positive and FH-negative screenees, respectively. This did not change at T2. There was no difference in risk perception in screenees who quitte smoking or lost weight versus those who did not.

Discussion
The main goal of genetic risk notification is to facilitate a free, well-informed choice for the client, reflected by an accurate risk perception, perceived effectiveness of preventive measures, and compliance with these measures. Less than 20% of the screenees rated their chance of having FH 1 in 2 (50%), the objective probability of first-degree family members of an FH-patient. Future FH-positive screenees rated their chance of having FH higher than the FH-negatives, only partly explained by the prior knowledge of cholesterol level. FH-positive screenees perceived a higher future heart attack rate than FH-negative screenees, but risk levels expressed numerically were again substantially lower than epidemiological risks in these patients. Screenees with a relative high risk perception more often used medication or opted for gene therapy. Also, medication utilization was the highest in screenees with the largest difference in risk perception between the situation with and without treatment. About 25% of the screenees smoked 7 months after screening, and, maybe even more disturbing, smoking status and BMI did not change over time nor differed between FH-positive and FH-negative screenees.

Some study features require consideration. A possible limitation of the study could be the sample size. Although our study included a large part of all Dutch screenees, it may not be large enough.
to conduct a detailed causal analysis in subgroups (defined according their initial attitude behavior or behavioral change). This research could be conducted in the future. However, the power of this study was large enough to show whether in FH-positive screenees the average changes aimed for were met and whether in FH-negative screenees adverse changes on average were absent. Another possible limitation is that all persons tested for FH were of Caucasian background, which could impair the generalizability of the findings of this study for other genetic screening programs. From a clinical point of view, validity seems unimpaired in our case, as FH-mutations in the Netherlands are primarily restricted to Caucasians. However, our results may not fully apply to screening programs involving all ethnicities, or with altered risk conditions in ethnic minorities.

The Dutch program did not include (co)payment of the screenees, either for the test or for subsequent treatment. To some extent this enables to investigate personal health beliefs without interference of financial motives, on the other hand generalizability may be limited for countries with another financial arrangement.

We had to decide on relevant time anchors for measurement: distant enough from the risk notification to study stable, deliberate, views on personal health risk and preventive behavior, and approximate enough to be able to study causal reasoning of the screenee. From clinical experts and additional data we collected we know that the clinical management of FH-positive screenees often take some months to stabilize.

A theoretical study feature, which needs consideration, is the risk perception concept used. We deliberately chose the concept of 'conditional own risk', that is the risk as perceived by the screenee given a certain condition. This concept has the advantage above unconditional measures in that it is more closely linked to preventive behavior. Perceived risk predicts behavior as well as comparative risk appraisal.

We included both numerical and verbal risk ratings, because numerical measures of risk perception are claimed to be best for checking the accuracy of screenees' risk perception as influenced by risk notification, while verbal measures are found to be better predictors of people's behavior intentions. The latter was not confirmed in our study, rather we observed similar relations of both risk appraisals to preventive measurements. There is no indication that selective lost to follow-up was present and the respondents were well able to answer the risk perception survey-questions.

Risk notification is generally directed at facilitating risk-reducing behavior. Rogers [1975] suggested that three main stimuli could influence risk-reducing behavior: perceived severity of the disease, perceived vulnerability and the efficacy of the recommended response. Others have recognized that perceived vulnerability in the sense of an accurate risk perception is probably a necessary prerequisite for behavior change.

In this study we concentrated on perceived vulnerability after risk notification. This study showed that the screenees had an inaccurate low numeric risk perception on having FH and on getting a heart attack, although apparently differentiated between the two. The rather low numerical risk perception of the heart attack rate with FH, is consistent with the literature: large risks on events are in general underestimated, and risk judgements frequently show moderate correspondence to epidemiological findings. One explanation of this low risk perception is inadequate patient education. However, other factors have to be taken into account. Firstly, unrealistic optimism of one's own health and health risks is a well-known phenomenon. Secondly, this study shows, as other studies, that screenees without a direct or indirect risk-experience have a less...
accurate risk perception than screenees with risk-experience. [Sjöberg, 2000; Weinstein, 1987; Wilcox and Stefanick, 1999] Apart from risk-experience, the age of the screenee compared to the affected family members could be of importance; the closer in age, the more likely a person might see him or her self at risk. [Hunt et al., 2001] Lastly, in this study we did not study the screenees’ prior expectations of and knowledge about the genetic test and diagnosis. These expectations and this knowledge could influence the way people handle the risk notification. However, unbiased measurement of the pretest status usually is impossible in practice due to (understandable) ethical requirements, which impose the candidate screenee to be informed on purpose of the screening offer and on preceding measurements.

Overall, the results show that the current information provision on vulnerability (having FH and getting a heart attack) fails to achieve a medical correct risk perception in the screenees.

Most models of preventive health behavior include the recognition of one’s own risk status as an important condition to make a rational decision on adopting risk-reducing behavior. [van der Pligt, 1998] This study shows that screenees with the highest objective risk and risk perception also have the highest medication utilization and more often opt for gene therapy when this will become available. The conclusion that risk-information suffices to enhance rational behavior must be tempered if we take smoking habits and body weight into account. The screenees seem to make a rational choice concerning medical treatment, but not concerning life style adjustments. This could indicate that FH is seen as a ‘medical’ problem, without a direct link to ‘non-medical’ life style factors. In general, people seem to be quite aware of the relative health risks of specific behaviors, but the interpretation of these risks changes when this knowledge is applied to own risk behavior. [Ayanian and Cleary, 1999; Lee, 1989; McKenna et al., 1993; van der Pligt, 1998] Also, screenees might only focus on what they feel is the most risky factor (in this case FH), and using the information about other ‘less relevant’ risk factors much less. [French et al., 2000] As smoking cessation might have the same or even larger health impact than reducing everyone’s cholesterol levels to normal [Bonneux, 2000; Tsevat et al., 1991], this casts doubts on the utility of FH-screening in the present form. It may be that too little attention is given to other risk factors than the genetic risk factor. However, risk-modification of the old risks presumably requires more investments than that of the new one. Furthermore, using medication or opting for something ‘futuristic’ like gene therapy demands less of the patients (or is perceived to be less demanding) than quitting smoking or changing one’s diet.

We conclude that the FH-positive screenees correctly perceive a higher risk of getting a heart attack later in life than do FH-negative screenees. Also, FH-positive screenees do not exaggerate their risks or feel that CVD are inevitable. Finally, risk perception was associated with medication use and the intention to opt for gene therapy, but not with other preventive measures, in particular smoking cessation. Thus, genetic risk notification seems acceptable and does not lead to aversion to preventive behavior. Also, correct representation of numerical risks is not a prerequisite for correct risk perception used for deciding on preventive behavior.
Acknowledgements

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