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Common chronic diseases (e.g. cardiovascular diseases, and type 2 diabetes) are becoming more prevalent due to growing physical inactivity and unhealthy eating patterns, resulting in an overweight population. A growing number of genetic variants that contribute to the multifactorial aetiology of these diseases are currently being identified. Testing based on these genetic variants alone (DNA-based test), or in addition to traditional disease risk factors, such as obesity and hypertension, still shows limited predictive value for disease. In the meantime, family history might be used as a ‘genomic’ tool for disease prevention and health promotion. Having a family history of these common diseases reflects the complex interaction of genetic susceptibility, common behaviours and shared environment, and numerous studies show that familial risk is an important and independent risk factor for these diseases. In this thesis, type 2 diabetes was taken as an example to explore the role of family history information in common disease prevention.

The central objectives in this study project were to examine (1) the impact of assessing and communicating familial risk of diabetes on risk awareness and behaviour change and (2) the possible large-scale positive and negative outcomes of integrating such a family history tool into public health.

Chapter 2 describes semi-structured interviews exploring the perceptions of causes, risk, and control with regard to diabetes and the role of family history among people at increased risk for type 2 diabetes. Individuals often mentioned a combination of genetic and behavioural factors as causes for diabetes. Some participants with a family history expressed incoherent causal beliefs; their general ideas about the causes of diabetes did not explain why their relatives were affected. The role of genetics as a cause for diabetes was more pronounced when people perceived diabetes as “running in the family” and this finding did not necessarily relate to a high number of affected relatives. Although people with a family history were aware of the diabetes in their family, they did not always associate their family history with increased risk, nor did they worry about getting diabetes. The absence of diabetes in the family was often used as a reason to perceive a low risk. Participants who primarily perceived genetic predisposition as a cause felt less able to prevent getting diabetes. Thus, future diabetes prevention strategies would benefit from giving more attention to individual perceptions, especially in the context of family history, explaining the multifactorial character of diabetes, and highlighting effective ways to reduce the risk.

Lay perceptions of issues related to predictive testing for diabetes, either using a family history assessment or DNA-based testing, were identified with focus group interviews in Chapter 3. Most participants believed in the ability of a family history assessment to identify people at risk for diabetes and to motivate preventive behaviour. However, a diabetes family history assessment was not considered useful by some participants, since there are also other risk factors involved; not everyone has a diabetes family history or knows their family history; and it might have a negative influence on family relations. Different reasons underlying motivation to change behaviour were considered when comparing a disease risk assessment based on DNA test results to a family history risk assessment. One perceived drawback of DNA testing was that diabetes was considered not severe enough for this type of risk assessment. Respect for the autonomy of individuals was not perceived to be an issue. Psychological harm, discrimination, and privacy were only briefly mentioned. In conclusion, these results show that laypeople believe that a predictive genetic test can be used in the prevention of diabetes, but indicate points to consider before both these tests are applied. These considerations differed with regard to the method of assessment (DNA test or obtaining family history) and also differ from monogenic disorders.
In Chapter 4, data from two randomized controlled trials, one in the United Kingdom (ADDFAM trial) and one in the Netherlands (PreDict trial), was used to assess the clinical value of using family history information for both coronary heart disease and diabetes, respectively. Findings showed that compared with a detailed questionnaire, a simple enquiry (i.e. a single question addressing family history) correctly identified the majority of individuals classified as having no significant family history, but missed a significant proportion of individuals with a positive family history. Incorrect classification of family history, in particular the high false-negative rate, has implications for the utility of a simple enquiry in identifying familial risk in clinical practice.

Chapter 5 describes a randomized controlled trial that assessed the potential effectiveness of communicating familial risk of diabetes in a personal consultation on illness perceptions and self-reported behavioural outcomes. Compared with individuals receiving general risk information, those receiving familial risk information perceived heredity to be a more important cause of diabetes, perceived greater control over preventing diabetes, and reported having eaten more healthily. Communicating familial risk increased personal control and, thus, did not result in fatalism. Although the intervention did not influence intentions to change behaviour, there was some evidence to suggest it increases healthy behaviour.

Chapter 6 reports a randomized controlled trial (PreDict trial) that examined the impact of web-based familial risk information on illness and risk perceptions and risk-reducing behaviour of people with and without a family history of diabetes. It showed that web-based diabetic familial risk information reduced worry related to diabetes risk and decreased dietary fat intake in low socioeconomic groups, but did not improve risk-reducing behaviour in the total population of people with a family history. Furthermore, the information did not significantly reduce risk-reducing behaviour and risk perception among individuals without a diabetic family history. Therefore, there is no evidence that familial risk information leads to false reassurance. The use of a detailed family history questionnaire resulted in a greater percentage of individuals reporting familial risk, and thus in a higher number of people being identified as having a high risk of getting diabetes.

Chapter 7 describes the perceived value of diabetic familial risk assessment and information for users of the web-based study (PreDict trial), and the perceived implications for individuals and families. It was shown that users perceive a general diabetes risk assessment with detailed familial risk information as valuable compared to a simple enquiry without being too time-consuming. Although many individuals perceived fear of being discriminated by insurance companies on the basis of their test results, few users of both a detailed familial risk assessment and a simple enquiry perceived a negative influence on autonomy, responsibility, and medicalisation.

In the final chapter of this thesis, the main findings are discussed. Overall, the study findings suggest that a detailed familial risk assessment improves diabetes risk identification, as more individuals report having a family history. Also, people do not evaluate this type of assessment as being more time-consuming than a simple enquiry. Familial risk information may lead to improved self-reported risk-reducing behaviour, especially among lower socioeconomic groups and when communicated in a personal consultation. After being informed about their familial risk, individuals report no negative effects on psychological well-being. Furthermore, they still believe that ways of preventing them from getting diabetes exist, suggesting there is no sense of fatalism. Moreover, individuals without a
diabetes family history had no difference in their risk-reducing behaviour or perception of their risk when compared to people who just received general diabetes risk information, and thus there was no evidence of false reassurance. In conclusion, the findings of this thesis show that family history is a promising tool in public health for common chronic disease prevention, such as type 2 diabetes.