

Attitudes towards genetic diagnosis and coping strategies of persons suffering from hereditary spastic paraplegia



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Introduction

The hereditary spastic paraplegias (HSP) are a group of late-onset, genetically caused, neurodegenerative disorders that can appear at any age. Both the clinical manifestations (with the main symptoms of a spastic gait disorder, muscular hypertonia and hyperreflexia) and the age of onset usually vary within the family and between families. The variation of the symptoms reflects the genetic heterogeneity of the disorders. For a small number of pure and complicated paraplegias, not only clinical, instrumental, laboratory diagnostics and imaging are available, but also genetic diagnosis is possible. With the availability of genetic diagnosis, the disease can be predicted in persons at risk and in unborn children (prenatal). This offers a new field for the investigation of psycho-social aspects in another neurodegenerative disorder. Our study continues earlier investigations carried out in families with other late-onset neurodegenerative disorders (Huntington's disease, Heredoataxias) and hereditary cancer disorders (hereditary breast and ovarian cancer, hereditary non-polyposis colorectal cancer syndrome). We aim to investigate the opinions and attitudes of families with spastic paraplegia towards genetic diagnosis, their coping strategies and the social and family situation.

Methods

The investigation was carried out by questionnaires (anonymous when requested) adapted from our previous studies and including the Trierer and Freiburger coping-scales. A random sample was interviewed personally or by phone.

We distributed 410 questionnaires as follows:

	Questionnaires delivered	Questionnaires received	Percentage
Patients	307	132	43.0
Patient's partner	38	12	31.6
Risk-persons	48	11	22.9
Risk-person's partner	17	2	11.8
Summary	410	157	38.3

Here, we report the results in the patients' group only:

Figures 1 to 4 deal with attitudes towards genetic diagnosis and the reasons for and against it.

Figure 5 shows the relationship of agreement to genetic diagnosis, actual engagement in it and the result of being a mutation carrier.

Figure 6 deals with the meaning of psychological support and figure 7 shows the coping strategies in the three groups: 1) not taking genetic diagnosis, 2) being a mutation carrier and 3) a non-mutation carrier.

The use and importance of some institutions and professional/non-professional persons are shown in figures 8 and 9.

Discussion

Unlike persons with other late-onset neurodegenerative disorders, families with HSP partners and persons at risk are less interested in problems dealing with genetic diagnosis. The acceptance of genetic diagnosis between affected persons suffering from HSP is very high; however, only half were actually undergoing genetic diagnosis. The main reasons for undergoing genetic diagnosis are the same as those known from earlier studies. However, the main problems against undertaking genetic diagnosis, such as psychological problems with regard to coping with the result and problems with the protection of data or with insurance companies are not seen in HSP. The coping strategies are not different between the three groups of not diagnosed, mutation carriers or not mutation carriers. As in other studies, active coping and the upgrading of self-esteem are the most important strategies. Because there are two self-help organizations with disagreeing views in Germany, they do not play such an important role as the German Huntington's or Heredoataxia Associations. Most of the patients suffering from HSP were diagnosed only by neurologists and were unaware of the possibilities of genetic counselling. Hence, GPs, neurologists, family members and partners were seen as being the most important possible partners in coping. Interestingly, more than 80% wanted to speak by phone about their problems; this reflects the great requirement for psychological support.

Conclusions

In Germany, patients suffering from HSP do not perceive problems in connection with genetic diagnosis as are known from other late-onset neurodegenerative disorders. Self-help organizations, GPs and neurologists do not deal with these issues and thus better co-operation is required between them, psychologists and genetic counsellors. However, patients suffering from HSP cope in a more active manner than those with previously studied hereditary diseases.

Acknowledgement

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Results

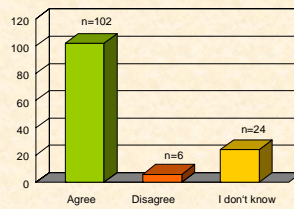


Figure 1: Opinion towards genetic diagnosis

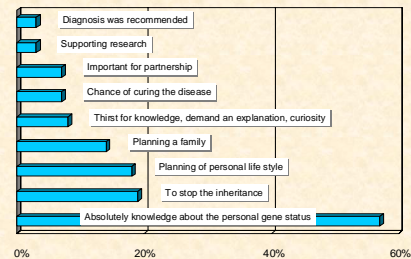


Figure 2: Main reasons for genetic diagnosis

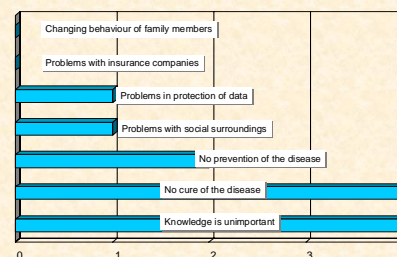


Figure 3: Main reasons against genetic diagnosis

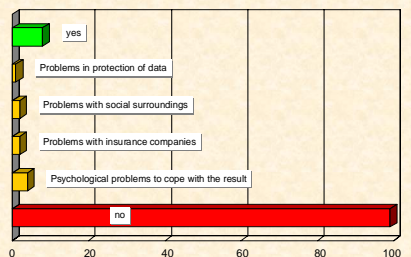


Figure 4: Do you see any problems in undertaking genetic diagnosis?

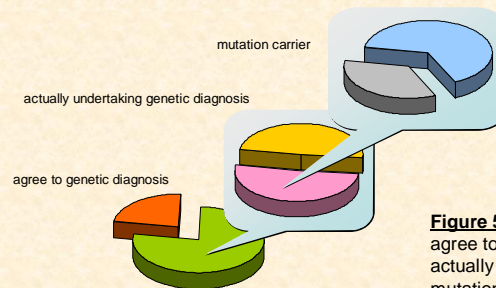


Figure 5: agree to genetic diagnosis actually undertaking genetic diagnosis mutation carrier

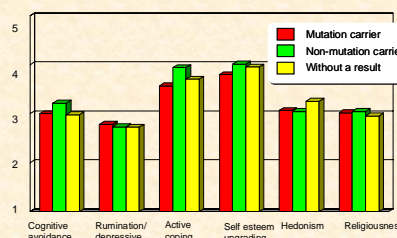


Figure 6: psychological support

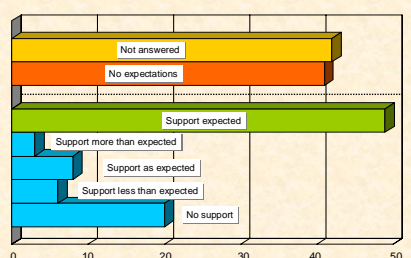


Figure 7: Coping

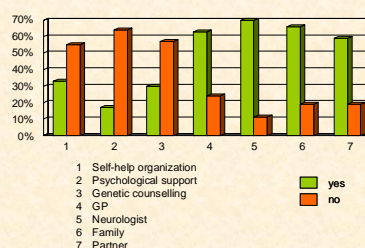


Figure 8: Use of ...

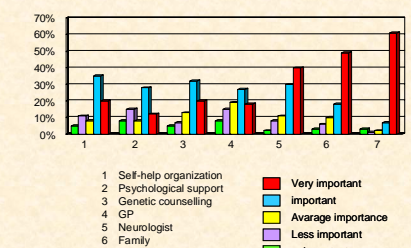


Figure 9: Importance of ...