Examining low-threshold support and guidance in rare genetic diseases

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Summary

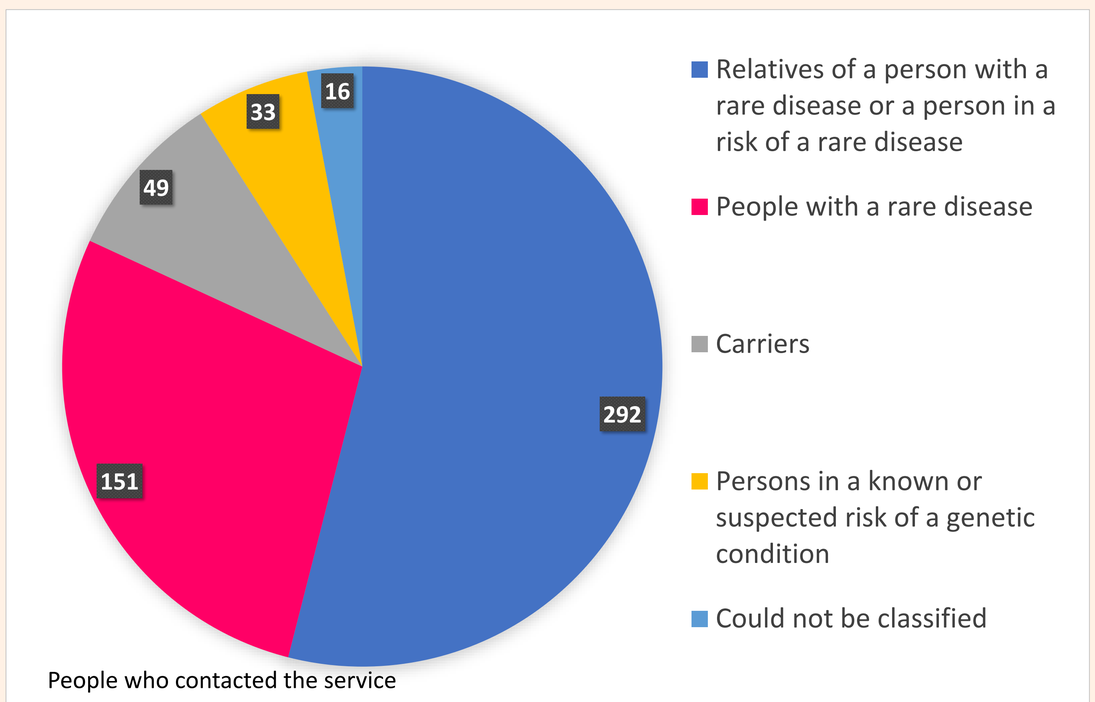
- Objective: to examine in what kinds of situations people want low-threshold support or guidance concerning genetics, inheritance and rare diseases, and to understand why the low-threshold service is used.
- Method: a review of the contacts made to a genetic counsellor during 2017
- Conclusions: People with rare diseases in their families, especially parents of children with rare diseases, wish to have more information about the disease and to discuss their fears and other emotions. The needs for the discussion vary depending on the life situation and on how much information there already is. The extra value of the low-threshold service, compared to other services, is the expertise of the genetic counsellor, who, unlike many other healthcare professionals, knows exactly what is being discussed.
- Significance: Recognizing the needs of people to discuss genetics, inheritance and rare diseases provides information about the services needed, as more and more genetic information will be available.

Background

About 80 % of rare diseases are genetic, and gene tests are available for ever more conditions. As genetic awareness is raising, people have all the time more questions concerning the inheritance of the diseases in their families, their risk and the risk of their children, as well as living with these conditions. Although people often receive appropriate information in public healthcare services, they still hunger for additional information, support and understanding. In Norio Centre, a Finnish centre for rare and genetic diseases, low-threshold support and guidance is offered to everyone interested by a genetic counsellor via telephone and e-mail or face to face. In 2017, 540 non-professional people contacted the service in order to ask about issues related to rare genetic diseases.

Method

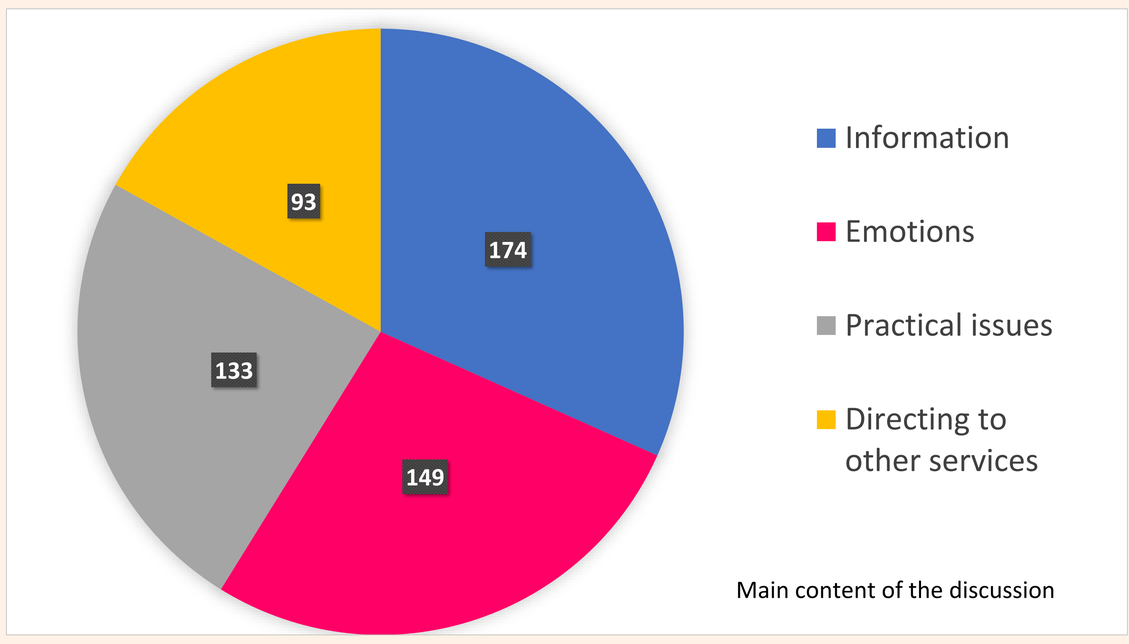
We studied all contacts with the genetic counsellor made during the year 2017. They were registered in a table summarizing the question asked, the condition or the situation in question, the duration of the contact, the conclusion made, and the feedback given. All this data was reviewed, categorized and analyzed at the end of the year.



People who contacted the service

Results

54 % of the persons who contacted the genetic counsellor were relatives of a person with a rare disease or a person in a risk of a rare disease. Most of them were mothers of small children with rare conditions. 49 % of the relatives wanted expert information concerning the disease and/or its inheritance. 34 % of these contacts were made due to practical reasons, e.g. to set a new appointment, and 31 % dealt with emotions, fears or anxiety related to the situation. 21 % of the relatives were guided to further services. The second biggest group, 28 % of the persons who contacted the service, were people who themselves had a rare disease. Their reasons to contact the service were otherwise like the reasons of the relatives, but only 21 % of them wanted information concerning the disease or its inheritance. The rest of the contacts were made by carriers or persons who were suspected to be carriers of a genetic condition (9 %) and by persons in a known or a suspected risk of a genetic condition (6 %). Most of these contacts dealt with information about the risk, inheritance, genetic testing and the impacts of the risk. 3 % of the contacts could not be classified. Most of the persons who contacted the service were very thankful for all information received and for the possibility to discuss their specific situation with a professional who understood what was being talked about.



Main content of the discussion

Conclusions

Low-threshold support is much needed in situations where there is a rare condition, or a risk of a rare condition in the family, and not much information or support is available in healthcare services or by other people. Especially mothers of children with rare diseases have many questions relating to inheritance, family planning, testing and treatment. They also often want to discuss their emotions. The needs of relatives, people with rare diseases, carriers and people in risk also vary. E.g. people with rare diseases often already have a lot of information about the condition and, thus, wish to discuss more specific issues. Although there are not always answers to all questions, many people are very thankful for all understanding and information. As there will be more genetic information available, there is a growing need for the interpretation of this information and support on the low-threshold basis.