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Individuals with different rare diseases are united by similar pre-diagnostic characteristics. A key to find the diagnosis earlier?

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Background. Individuals with a rare disease (RD) are facing many different challenges, but a common feature for many of them is the pre-diagnostic odyssey. We hypothesized that individuals with different RDs share distinctive features during the pre-diagnostic time. These features could be used for diagnostic support.

Patients and Methods. In a Delphi survey among German experts, 20 RDs with utmost need for diagnostic support were identified. Interviews with 10 adults with a RD and 10 parents of children with a RD were performed, transcripted and qualitatively analyzed using the method described by Colaizzi. Using this stepwise approach categories describing pre-diagnostic peculiarities were verbalized and a specific and unique questionnaire created.

Citation	Thematic group	Question
"I spoke to my dentist, I say: "This looks so weird, or my teeth no longer fit over each other", and the reaction was then a shrug - "yes". Yes. Then that was it." *(10/68-71).*	Search for causes **Category** Presented symptoms remain without consequence	Is it true that your ailments/ irritating appearances by doctors/ your doctor remained without reaction at first?

Table 1. stepwise creation of a questionnaire

Results. A variety of different pre-diagnostic features were identified during the analytic process. The final questionnaire contains 53 questions (e.g. 'Did you perform research regarding possible causes for your symptoms?', 'Did you experience a key moment that made you feel threatened by your symptoms?'). With the help of patient organizations the questionnaire was completed by more than 1.000 individual with a RD.

Instructions for usage of the diagnostic tool. An individual without diagnosis can answer the questionnaire. Data mining methods will then compare this answer pattern with those in the data base. A categorisation into one of 6 groups is possible today: 1. Rare disease; 2. Chronic disease; 3.healthy; 4 psychosomatic disease; 5. 'no diagnosis'; 6. 'other'. The final diagnosis depends on further tests.

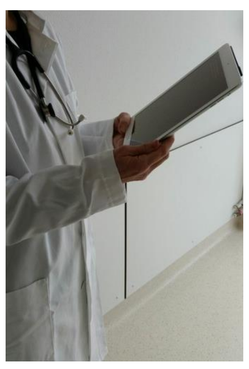
Category	Disease	Replies (n)
Rare Disease	***e.g. sarcoidosis, pulmonary hypertension, neuromuscular diseases***	**743**
Chronic Disease	***e.g. fibromyalgia; inflammatory bowel disease***	**174**
Somatoform Disorders	***Diagnosis confirmed in tertiary hospital***	**34**
No Diagnosis	***No diagnosis after extensive diagnostic work-up***	**34**
Healthy	***Individuals who categorize themselves as healthy***	**22**

Table 1. More than 1000 individuals with rare diseases, chronic diseases, psychosomatic diseases answered the questionnaire. A large data base was built. Examples are given in table 1.

Discussion. Individuals with a RD share common features during the pre-diagnostic time. A questionnaire-based diagnostic support tool was developed. This tool – together with data mining methods – can be used to increase the awareness for a RD. Certain questions have the potential to serve as 'red flags' to identify an individual with a RD. It remains open whether trans-linguistic and trans-cultural transfer of a questionnaire based tool is possible.

QR code to access the german questionnaire

Patients without diagnosis are sometimes lost in a „diagnostic labyrinth"



Our questionnaire-based tool can be used by physicians to rule out a rare disease

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