

Modelling Family Outcomes: from the Iceberg Syndrome to the Family Ecosystem, a Call for Innovative Patient-Driven Research



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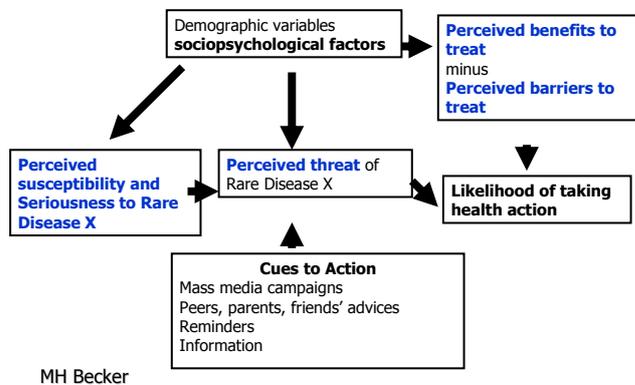
Introduction

The European Project for Rare Diseases National Plans Development (EUROPLAN) has addressed one unanswered question to date: "what is the scope of patient-driven research?" Identification of needs and priorities for translational and social research, modes of fostering them and promotion of interdisciplinary cooperative approaches has been largely recommended. Beside the success stories of "orphan drug development" and genetic research during the last decades, family story telling and family sketches analysis show minor progress in the clinical management and day to day care despite tremendous unmet needs. Quality of Life and measurements of the burden of the disease have been neglected. A call to action for active research in the field of human and social sciences is urgent.

Modelling

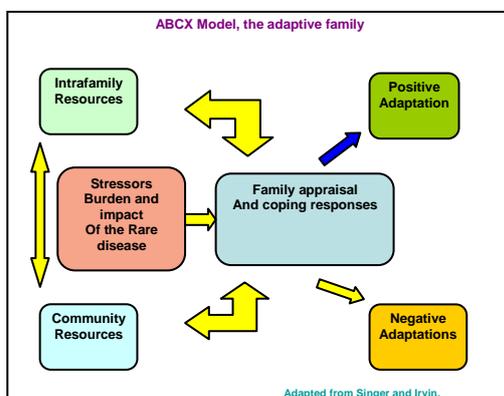
1) Rare Diseases obey to the **social model of health beliefs**

The lack of curative treatment is not a barrier to adopt a health action.



MH Becker

2) Taking health action = **adaptive strategy**

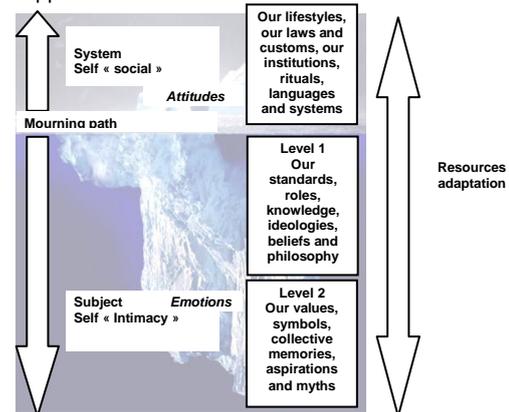


Adapted from Singer and Irvin,

Day to day life with a Rare Disease could be seen as an adaptive strategy using a stress coping model. The stressors here are the family crisis.

3) Attitudes and beliefs are included in the **Iceberg Syndrome**

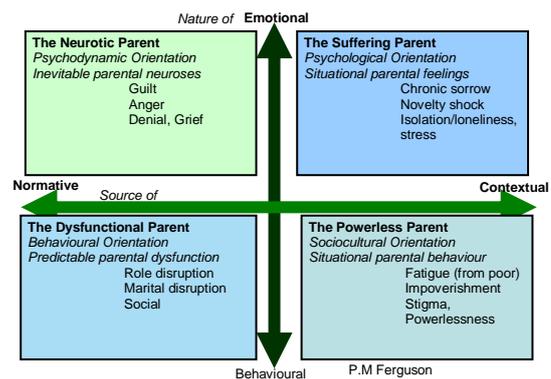
Metaphorically the Rare Diseases history is like an Iceberg with a 10% visible part and 90% of unanswered solutions for a better quality of life or at least solutions to support the burden of the disease.



The Iceberg Syndrome model provides a systemic approach of the burden of a Rare Disease, therefore an overview of patient outcomes and a model of resource adaptation.

4) Family portraits

Portraits could be classified in a matrix with a vertical axis from behavior to emotion for the impact and from the family unit to the environment for the source of the problem.



Conclusion

Models provide pathways for a better understanding of needs and also a spectrum of Patient-Driven Research perspectives.

Research including Sociopsychological projects will provide cues to responses to so many day to day unmet needs and suffer beside the need to have a speculative curative therapy. Patient and family outcomes in the field of Orphan Drug development and Research in Rare Diseases should be systematically assessed to respond to Public Health Priority.