

Growing up with a rare condition

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Introduction

The WHO International Classification of Functioning, Disability and Health (ICF) can be used to describe a person's health state in detail using a bio-psycho-social perspective. The central concept of the ICF is the concept of functioning. Together, WHO standards ICF, International Classification of Disease, and the Anatomical Therapeutic Chemical are classification to describe and relate medical conditions, medicines, health and well-being in digital health records [1]. Shwachman Diamond Syndrome (SDS) is a genetic condition apparent from childhood, characterized by pancreas insufficiency, neutropenia, skeletal anomalies and autism-like behavior. The aim of the pilot study was to correlate the ICF classification with personal experience of adults with SDS.

Materials & Methods

Representatives of the Dutch SDS patient organisation selected 12 categories from the domain activities and participation of the ICF core-set autism brief [2] and included these items in a questionnaire. The five participating adults with SDS responded to the questionnaire in writing (scoring the items and adding additional remarks) and subsequently they underwent a structured interview. Two ICF experts analysed the additional remarks and the reports of the interviews using AtlasTi version 22. The quotations selected were scored with two level ICF codes. ICF codes used at least six times in three or more respondents are included in the table.

Activities and participation	Functions
d920 Recreation & leisure	b152 Emotional functions
d240 Handling stress and other psychological demands	b126 Temperament and personality functions
d850 Remunerative employment	b455 Exercise tolerance functions
d570 Looking after one's health	b125 Dispositions and intra-personal functions
d475 Driving	
d310 Understand spoken messages	Environmental factors
d720 Complex interpersonal interactions	e310 Immediate family
d610 Acquiring a place to live	e330 Peoples in positions of authority
d750 Informal social relationships	e355 Health professionals
d640 Doing housework	e360 Other professionals
d710 Basic interpersonal interactions	e120 Transportation
d230 Carrying out daily routine	
d210 Undertaking a single task	

Results

The table shows ICF ≥ 6 quotations from ≥ 3 respondents; the most frequent are on top. Not only activities and participation categories were used frequently, but also functions and environmental factors.

Conclusion

Understanding the positive / neutral and negative aspects of living with a rare condition may help parents and communities to support growing up towards a fulfilled life. Incorporating the ICF in personal digital health records promotes health and well-being at all ages (Sustainable Development Goal #3, United Nations) [3].