

# Living with Shwachman Diamond Syndrome

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## Introduction

The International Classification of Functioning, Disability and Health (ICF) and the International Statistical Classification of Diseases and Related Health (ICD) ensure semantic interoperability of data in life.

Shwachman Diamond Syndrome (SDS) is a ribosomopathy resulting in neutropenia, pancreas insufficiency, metaphyseal chondrodysplasia and developmental disabilities. Diagnosis is made on clinical grounds and confirmed with mutation analysis.

The ICF-d chapter facilitates individuals to score the possibilities. The nine ICF-d chapters of the domain activities and participation describe the execution of a task or action by an individual, and the involvement in the life situation. The aim of the pilot study was to correlate the ICF classification with personal experience of adults with SDS.

## Patients and Methods

The Dutch SDS patient organisation selected 12 ICF-d – terms from the validated ICF autism core set (1). The questionnaire was sent to adults with SDS known to the patient organisation. 5/9 responded. In addition all participants were semi structured interviewed.

## Results

All responses were evaluated on the frequency and levels of problems registered: 0= no difficulty; 1= mild difficulty, 2= moderate difficulty, 3= severe difficulty, 4= complete difficulty.

Table number of responders with a score of **.2 (moderate difficulty) or more, without help.**

An individual digital interoperable ICF qualifier would be for example 640.2: meaning moderate difficulties with doing housework.

|   |          |
|---|----------|
| <b>820 School education</b>                               | <b>5</b> |
| <b>720 Complex interpersonal interactions</b>             | <b>4</b> |
| <b>920 Recreation and leisure</b>                         | <b>4</b> |
| <b>210 Undertaking a single task</b>                      | <b>3</b> |
| <b>240 Handling stress and other psychologic demands</b>  | <b>3</b> |
| <b>610 Acquiring a place to live</b>                      | <b>3</b> |
| <b>640 Doing housework</b>                                | <b>3</b> |
| <b>850 Remunerative employment</b>                        | <b>3</b> |
| <b>310 Communication with- receiving- spoken messages</b> | <b>1</b> |
| <b>4751 Driving a motorized vehicle</b>                   | <b>1</b> |
| <b>570 Looking after one's health</b>                     | <b>1</b> |
| <b>710 Basic interpersonal interactions</b>               | <b>1</b> |

1. ICF CORE SETS Manual for Clinical Practice  
Bickenbach J, Cieza A, Selb M, Stucki G (eds)  
Hogrefe, Göttingen 2020

## Quotes from the interviews

*'Why am I different'*

*'Everything new is difficult'*

*'I can not do two things at the same time'*

*'I am often ill and tired and there is little compassion'*

*'My communication is not so good'*

*'I live my life like I want'*

## Conclusion

Experiences during school education still play a major role in the life of people with SDS.

Effects of new therapies, medical aids and social support on daily life can be graded over time by the individual.

The individually scored qualifiers can be stored in a secured personal health environment and shared with health care providers and social institutions.

Additionally anonymized data on personal performance in daily life due to a genetic diagnosis can be associated with standards as the ICD.

Thus the ICF provides a valuable tool for healthcare providers, researchers, and policymakers, as it allows for the comparison of health and disability across different populations and the tracking of changes over time.

ICF is widely used in a variety of settings, including clinical practice, rehabilitation, and public health, and has been translated into many different languages for use around the world.

Stichting Shwachman syndroom

Support Holland



Persons with SDS, family, care takers, healthcare providers, researchers, and policymakers

You can all join and try the SDS ICF questionnaire.  
The answers are anonymous.

[https://rarecare.inventable.lk/pt\\_index.php](https://rarecare.inventable.lk/pt_index.php)

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