UNIVERSAL DATA COLLECTIONS, INCLUDING THE ICF

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Introduction

UNICEF states that millions of children with disabilities around the globe continue to be left behind. Often, this neglect is the result of limited data.

Over 70 percent of rare conditions have a genetic cause and manifest in childhood. One genetic defect usually causes a variety of disease manifestations, coded with the International Classification of Disease.

We questioned adult persons with Shwachman Diamond Syndrome (SDS) using ICF terminology on activities and participation.

Patients and methods

SDS is a rare ribosomopathy manifesting with pancreas insufficiency, metaphyseal dysplasia, developmental disability and neutropenia before age of 5 years.

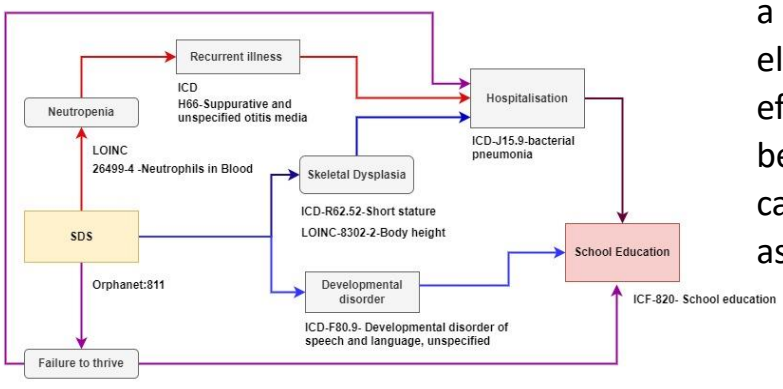
Children are hospitalized for bacterial infections, failure to thrive and skeletal dysplasia.

We selected 12 ICF-d domains from the short autism core set affecting SDS. 5/9 adults with SDS responded to the adjusted SDS core set.

Results

Table number of responders with a score of .2 or more, with or without support, as recorded in the ICF Questionnaire.

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



Conclusion

Lack of appropriate school education has a major impact on adults with SDS. The developmental disorder, recurrent illness and hospitalisation can all have had effect on school education. Therefore we suggest the ICF to be associated with a rare disease coding capturing the ICD registrations. To ensure no one is left behind.

For families:
Using the ICF-d scores associated with a rare disease classification individuals with a rare condition can generate data in electronic health systems. Data on the effect of having a rare condition on well-being will become available. These data can be associated with clinical data such as the ICD.

You can all join and try the SDS ICF questionnaire. The answers are anonymous.
https://rarecare.trainable.lk/pt_index.php

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