

Down Syndrome Program

A conceptual model of health in Down Syndrome

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Background

The WHO defines health as “a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity.”¹

In the US, over 200,000 individuals with Down syndrome (DS).

What defines health for individuals with Down syndrome?

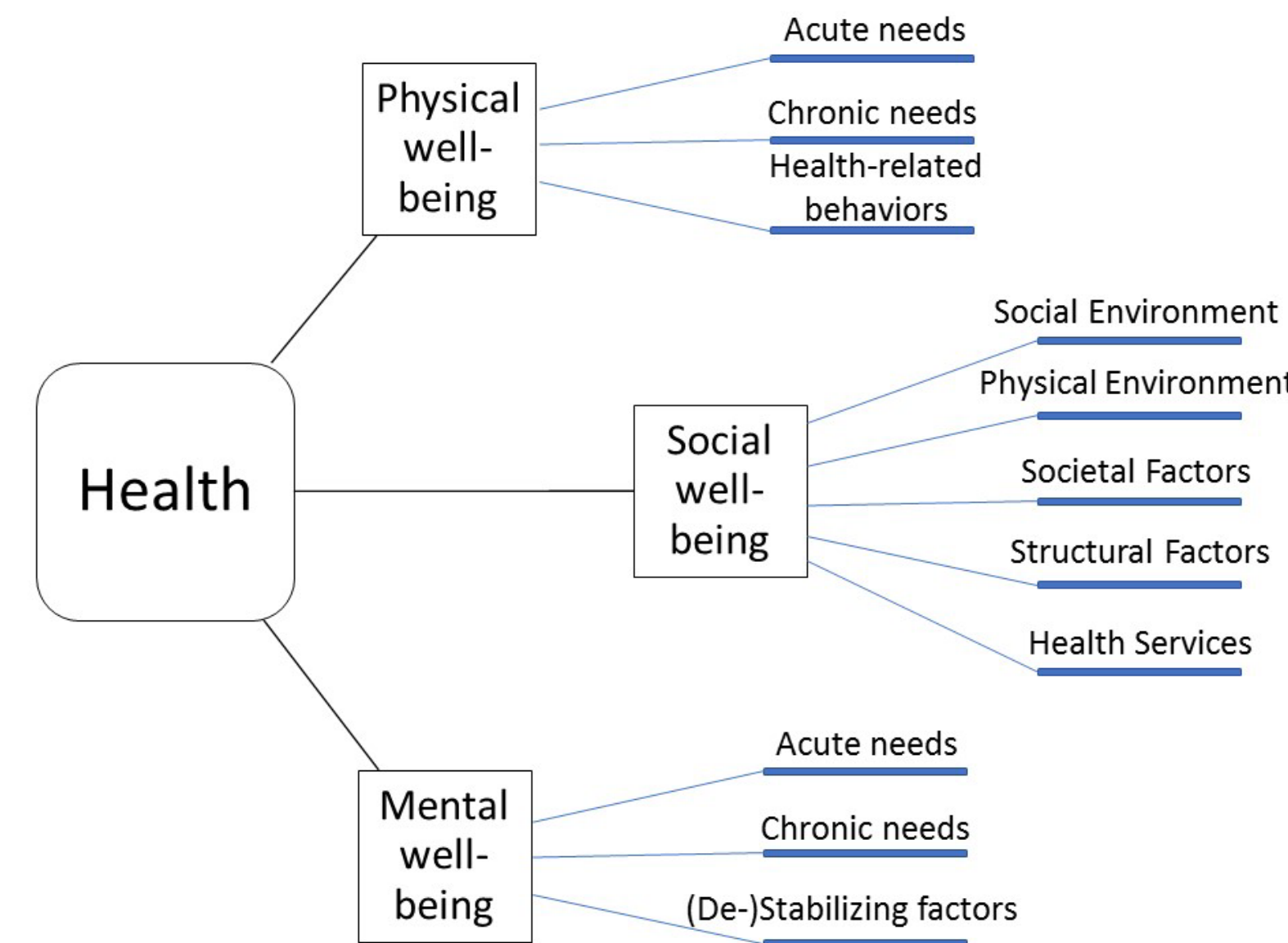


Figure 1: Preliminary conceptual model of health in Down syndrome based on WHO definition of health, and literature review

Is the definition of health similar to people without Down syndrome?
Is “health” similar to “health-related quality of life”?

What makes a day a healthy day?
How healthy do you feel?
What are signs of being in good health?

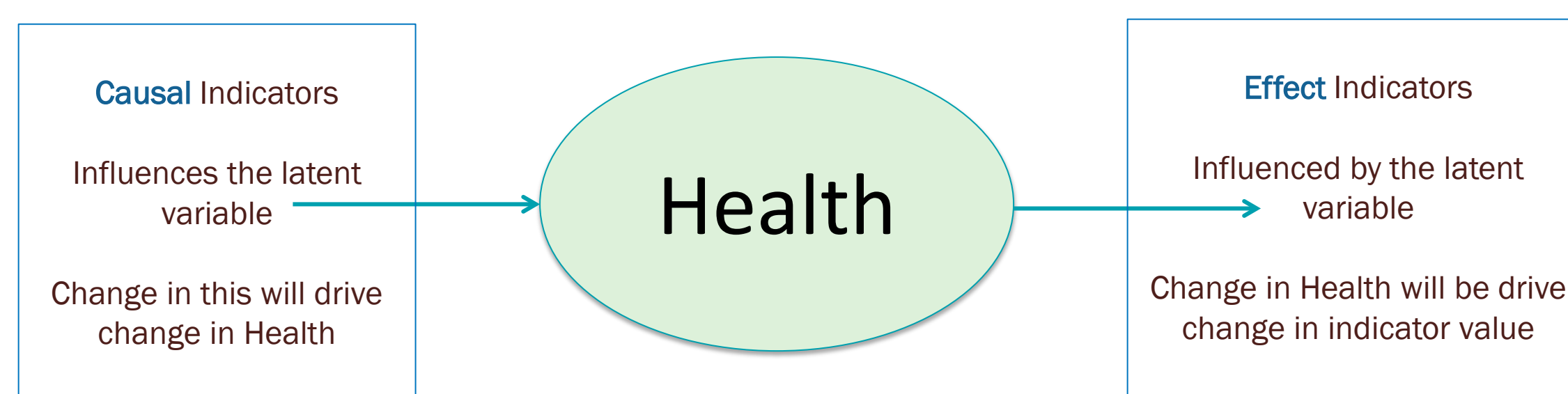


Figure 2: Example indicators (causal vs effect) of socioeconomic status

Methods

In 2021: 3 expert panels, 8 focus groups

- parents of individuals with Down syndrome age 0–21 years
- individuals with Down syndrome age 13-21 years were conducted through videoconferencing technology.

We showed preliminary model to participants, and discussed the concept of health.

Approved by MGH Institutional Review Board.

Results

Tables 1 & 2: Demographic features of participants in Expert Panels (N=23) and Focus Groups (N=28)

	N	Trait	Response	N
Physician	6	Persons with Down syndrome in Focus		
Psychologist	3	Groups		
Behavior Expert	1	Age (years)	13-17	4
Educator	5	Sex	Female	4
Legal Advocate	1	Parents in Focus Groups		
Advocate	1	My child with Down syndrome's age	0-5	5
Speech and Language Pathologist	1		6-12	4
Social Worker	1		13-17	5
Genetic Counselor	1	My child with Down syndrome's gender	18-21	6
Family Support Specialist	2		Male	10
Researcher	1		Female	10
		Parent Sex	Female	19
		Parent Race	White or Caucasian	17
			Black or African American	1
			Asian	1
			Other	1

Mental Health Constructs
Motivation
Identity
Quality of Life
Prioritize Tasks
Adaptive Behavior
Self-regulation of Behavior
Mental Health Symptoms
Behavior to Communicate

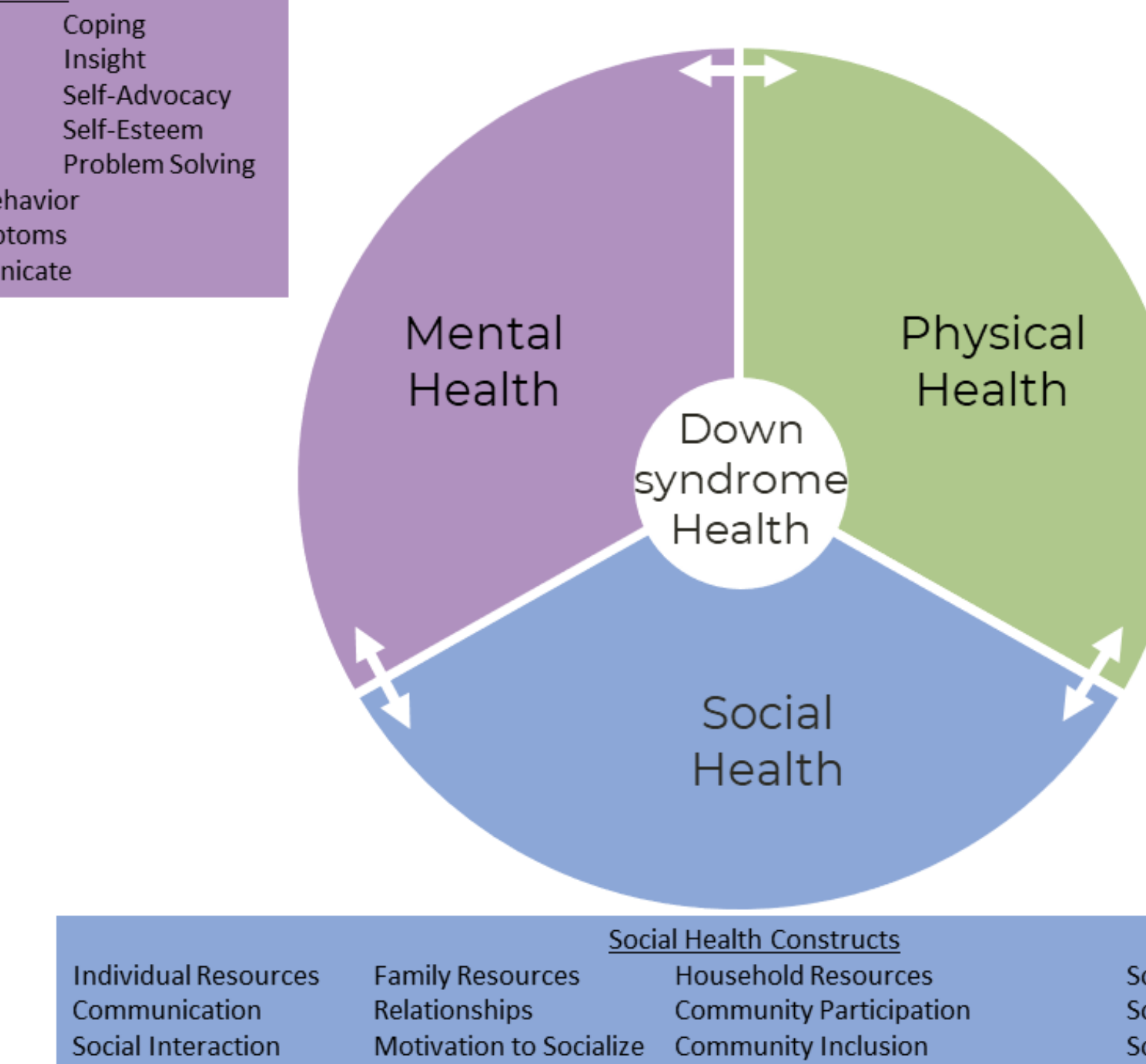


Figure 3: Final conceptual model of health in Down syndrome

Discussion

Experts and parents agreed that individuals with Down syndrome have unique health concerns that could be better understood using a conceptual model of health. Their unique health concerns led to specific constructs within each dimension and necessitated the creation and validation of a syndrome-specific health model for future research.

Future study will use this model to measure health in Down syndrome.

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Table 3: Causal and Effect Indicators of Physical Health in the Conceptual Model of Health in Down Syndrome

Each “+” indicates one focus group or expert panel during which the topic was discussed

Color coding: + = FG1, ++ = FG2, +++ = FG3, ++++ = FG4, +++++ = FG5, ++++++ = FG6, +++++++ = FG7, ++++++++ = FG8; ++ = EP1, +++ = EP2, ++++ = EP3

Individuals with DS Focus group (FG) endorsed (N=4)	Parent Focus group (FG) endorsed (N=4)	Expert panel (EP) endorsed (N=3)	Causal Indicators	Effect Indicators	Individuals with DS Focus group (FG) endorsed (N=4)	Parent Focus group (FG) endorsed (N=4)	Expert panel endorsed (N=3)
++++	++++	+++	Diet and nutrition	Activity	++++	++++	+++
++	++++	+++	Medical Diagnoses	Metabolism	++	++++	+++
++	++++	+	Medications (including vitamins)	Sleep	+	++++	+++
+	++++	++	Procedures and tests	Sensory organs	+	++++	++
+	++++	+	Medical devices (G-tube, trach, glasses, ear tubes, walker, wheelchair)	Dermatologic symptoms	+	++++	+
+	+++		Medical Emergencies	Neurologic symptoms	+	+++	++
	+++	++	Family History	Cardiovascular symptoms	+++	+	+
+	+++		Dental	Mobility / Motor function	+	+++	++
	+++		Immunity	Oromotor function	+	+++	+
				Respiratory symptoms	+	+++	
				Musculoskeletal symptoms	++	++	
				Genitourinary symptoms	+	++	
				Pain	++	++	++
				Digestive symptoms	++	++	+