Table 3. Patient Changes from the Framework for Outcomes in Clinical commUnication Services (FOCUS)

Patient Change Categories[a]	**Description**	**Example measures**	**Hypothesized relationships with other patient changes, patient experiences, or health outcomes**
Knowledge	Objective knowledge about health topics (e.g.): 1) the condition for which the person is at risk; 2) level of risks and options for oneself or one's family members; 3) benefits and limitations of the various options; 4) treatment or action plan and procedural knowledge (i.e., how to take action).	Proportion of individuals who can accurately recall information about the benefits and limitations of their genetic testing options. Proportion of patients who can accurately recall which relatives are at risk for a genetic condition. Knowledge scales specific to: HBOC[1]–[3]; Lynch syndrome [4];Thalassemia [5] Fragile X [6]; Maternal Serum Screening [7]	Knowledge is often necessary but may be insufficient for other patient changes (e.g., access to care, quality health decision, adherence, and communication with family members.)
[b]Feeling informed [8]–[16]	Belief that one has sufficient information about the personal and family implications of the health condition/risks as well as information about available options for managing, clarifying, or dealing with the condition/risks.	Proportion of individuals who feel like they have enough information about a medical condition. Cognitive Control- Genetic Counseling Outcomes Scale [10] Decisional Conflict Scale: Inverse of the "feeling uninformed" subscale Patient Decision aids: Decisional Conflict Scale [17] Validation of a decisional conflict scale [18]	Feeling informed is likely necessary but may be insufficient to facilitate other patient changes (e.g., access to care, quality health decision, adherence, and communication with family members.)
Perceptions of health risks	Genetic risk perceptions - patient beliefs about how likely it is that they have a genetic predisposition. Perceived disease susceptibility- perception about how likely they are to develop symptoms associated with a condition. Perceived severity - perception of "how bad' the condition seems.	Patient's perception about their risk for cancer (can be absolute risk or relative to a reference group) Susceptibility and severity scales have been created based on the Health Belief Model and Extended Parallel Process Model [19, 20]	Together perceived risks, susceptibility and severity create a perception of threat and this can motivate or hinder access to care, adherence or self-management. Individuals who can reframe a situation and recognize or

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	Overall appraisal of health risks, conditions, or situations as positive or negative.	Primary and secondary stress appraisals can be measured based on Lazarus and Folkman's Transactional Model of Stress and Coping Transactional theory and research on emotions and coping [21]	focus on positive aspects of a health risk or condition may experience fewer negative emotions and better mental health.
	Levels of uncertainty about the condition	Uncertainty in illness scale [22, 23]	Strategies to manage uncertainty can improve symptoms and may decrease negative emotions.
Beliefs, motivations, & emotions about: 1) a decision, 2) behavior, or 3) anticipated outcomes	Attitudes, emotions, motivation, and perceived barriers related to the behavior or decision Anticipated outcomes (i.e., behavioral expectancies, perceived benefits, response efficacy, anticipated emotions) Beliefs about what healthcare providers and significant others want the patient to do Perceived importance of the decision or behavior Confidence in one's ability to take action and overcome barriers to do so (i.e., self-efficacy).	Several example scale measures to assess changes in this category are available based on commonly used health models like the Health Belief Model; Theory of Planned Behavior; Transtheoretical model (decisional balance); Self-determination theory; Extended Parallel Process Model; Social Cognitive Theory [19, 20, 24–26]	Low perceived barriers as well as positive attitudes and emotions about both the behavior or decision and the expected outcomes may lead to a quality health decision and improve adherence/self-management. Intrinsic motivation and high perceived benefits increase the likelihood of long term adherence/self-management. If significant others and/or trusted healthcare providers are supportive of an action it may be more likely that the patient will take that action. Self-efficacy is often a very strong correlate of behavior.
[c]Empowered to make quality decisions [8, 14–16, 27, 28]	Feeling supported, confident in and control over one's ability to make decisions that: 1) will maximize health and well-being and; 2) are consistent with the values	Decisional Conflict Scale (DCS) measures uncertainty in decision making (opposite of empowered to make a decision). Patient Decision aids: Decisional Conflict Scale [17] Validation of a decisional conflict scale [18]	Decisional empowerment contributes to whether a quality health decision is made and adhered to.

	and interests of oneself and perhaps one's family.	SURE scale measures certainty in decision making, perception of understanding of risks and benefits, clarity of which risks/benefits matter, perceived level of support/advice [29] Decisional regulation sub-scale of the Genetic Counseling Outcomes Scale [10] Decisional control from perceived personal control scale [15] Self-regulation and competence scales based on Self Determination Theory [30] Proportion of patients who are clear about the role their values may play in making a decision	
[d]Empowered to access resources and/or engage in self-management [8, 10, 14, 15, 27, 28, 30]	Feeling supported, confident in, and control over one's ability to effectively access and use medical and social support resources and engage in self-management.	Behavioral control subscales from either the perceived personal control [15] or GC outcomes scale [10] Illness management self-efficacy scales Self-Efficacy for Managing Chronic Disease 6-Item Scale [32] Self-efficacy and Quality of Life among People with Bipolar Disorder [33] Perceived competence scales based on Self Determination Theory [30] PROMIS measures: Emotional support, informational support, and instrumental support – assesses availability of information, and availability of assistance with tasks [33–35] PROMIS measures: Self-efficacy for managing chronic condition [37]	This is expected to improve adherence, self-management, and effective coping.
[e]Empowered to cope with emotions and uncertainty [8, 10, 14, 27, 28]	Feeling supported, confident in and control over one's ability to cope by effectively managing emotions & dealing with uncertainty.	Emotional regulation subscale in GC outcomes scale [10] Emotional self-efficacy scale [38]	This is expected to improve effective coping and reduce negative emotions.

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Behavioral or decisional intention [38, 39]	Intention, desire, willingness to follow through with a decision or a particular action plan (e.g., proceed with testing).	Behavioral intention measures using 5 or 7-point Likert scale [41]	Although intentions and desires do not always lead to action, those who lack intention or are ambivalent are extremely unlikely to follow-through.
Quality health decision [11–13]	Informed decision about a health behavior or medical care is made based on best available evidence; the decision is concordant with patient values & goals; and the decision can be implemented.	Proportion of *BRCA* mutation carriers who elect to have a prophylactic oophorectomy, believe they are ready to do so, and feel like potential benefits outweigh potential harms. Proportion of individuals whose decisions match their reported values. Proportion of individuals who were able to act on their decision Multidimensional Model of Informed Choice [42] Decision regret scale [43]	Quality health decisions can increase survival, reduce symptoms, and contribute to other health outcomes. A person can make a quality health decision, but still regret that decision later. Regardless, decisional regret can negatively impact health.
Accessed appropriate care [11, 13, 30]	Patient receives appropriate follow-up care and/or testing. Patient receives services necessary to make appropriate transitions (e.g., adult health care, employment, long-term medical care).	Proportion of children with developmental delays who receive appropriate therapies as the result of a genetics visit or accurate diagnosis.	Appropriate care can increase survival, reduce symptoms, or improve quality of life.
Adherence and self-management [11–13]	The patient follows through with health recommendations, action plan, or decisions, taking ownership of and being proactive about their care.	Proportion of patients with PKU who maintain phenylalanine levels within the medically accepted range. Self-care Behavior Inventory (McLaughlin 1985) [44] Genetic Counselor involvement has been associated with increased adherence to provider recommendations [45]	Adherence and self-management can contribute to improvements in survival and other health outcomes.
Activated support resources [46]	Patient accesses or receives desired levels of social	Of those individuals who desire more instrumental, emotional or informational support, the proportion who	Patients who activate support resources may have

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	support, emotional support, or additional information related to the health issue/condition.	report accessing it as a result of their new diagnosis or health care received.	improved mental and social health.
Reduction in negative emotions	Patient reduces negative emotions that are related to the health threat or diagnosis (e.g., anxiety, distress, anger, stigma, guilt, shame, worry, feeling overwhelmed).	Impact of events scale (measures distress related to a certain event or threat) [47] Multidimensional Impact of Cancer Risk Assessment (MICRA) measures distress, uncertainty, and positive experience (the latter would fit within coping/adaptation). [48] Psychosocial Illness Impact (negative item bank) PROMIS	Reduction in these negative emotions can improve mental health.
Effective coping and adaptation [11–13, 46]	Patient uses coping mechanisms that are associated with better patient health outcomes. Over time a patient may even find a sense of purpose to the situation and/or they may experience personal growth or meaning from the health condition or threat.	Increase in the proportion of patients who use coping strategies that are helpful to them or promote positive health outcomes. Brief COPE Ways of coping scale (Lazarus) Ways of coping [50] Ways of coping questionnaire [51] Psychological Adaptation Scale (PAS) –measures extent to which condition has helped a patient grow or find meaning Psychosocial Illness Impact (positive item bank) PROMIS	Effective coping and finding meaning in the situation contribute to a positive sense of well-being.
Reduction in use of unnecessary or more costly services	Patient does not seek care that is unnecessary	Proportion of individuals who have a known mutation in the family who undergo single site testing rather than full gene sequencing. [As prices of testing go down though, this will not result in much cost savings.] Reduction in number of ER visits among patients with metabolic conditions. Reduction in unnecessary medical procedures among individuals found to be at low risk for disease based on genetic testing and/or family history assessment.	Needs for emergency medical services are minimized when patients adhere to treatment plans. Correct diagnoses and accurate knowledge may reduce unnecessary health services.

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a. Patient changes are factors that change as a direct or indirect result of health services received. These are expected to influence patient health outcomes and/or family changes.

b. This encompasses the concept of “understanding” from Pithara [8] and McAllister’s concept of “cognitive control” [27]. This term also encompasses aspects of “perceived personal control” [15]. Lastly, it is also the inverse of feeling uninformed, which is a component of “decisional conflict” [16].

c. Inclusive of Pithera’s concept of “informed and shared decision making”[8] as well as McAllister’s concept of “decisional regulation” [27]. This term also encompasses aspects of perceived personal control [15] and decisional self-efficacy [28]. Finally, this construct is the inverse of several components of “decisional conflict” [16].

d. Inclusive of Pithara’s concept of “enablement” [8] and McAllister’s concept “behavioral control” [27]. This term also encompasses aspects of “perceived personal control” [15] and “behavioral self-efficacy”[28].

e. Inclusive of Pithera’s concept “reassurance” [8] and McAllister’s concept “emotional regulation” [27]. It is also similar to “emotional self-efficacy” [28].

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