The Role of Patient Experience in Rare Disease Drug Development

Alison Skrinar, PhD
Vice President, Endpoint Development and Strategy
Ultragenyx

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What do we mean by “Patient Experience”?

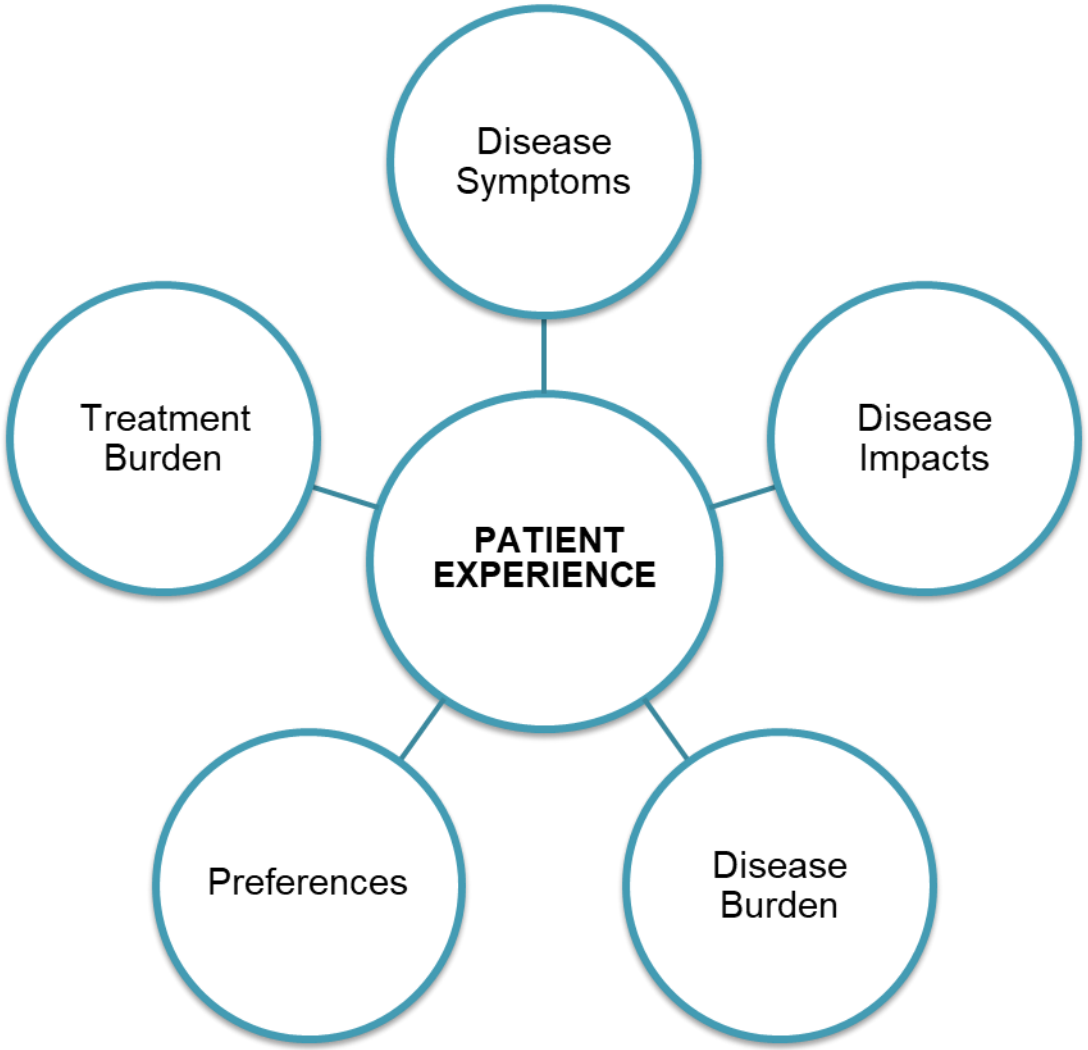
“Patient Experience” Reflects the Journey of Rare Disease

- Data that provide information about patients’ experiences with a disease or condition
  - Sources may include patient, caregivers, clinicians, therapists, etc.

- Includes the experiences, perspectives, needs and priorities of patients related to (but not limited to):
  - Symptoms of their condition and how they evolved over time
  - Impact of the conditions on daily functioning and quality of life
  - Experience with existing treatment and management regimens
  - Identification of most important outcomes for a disease-modifying treatment
  - Relative importance of identified outcomes from perspective of patients
  - Patient preferences for treatment (frequency, administration route, side effects)

Source: Title III, Section 3002(c) of the 21st Century Cures Act

Components of Patient Experience Data



Why is Patient Experience Data Important?

- Patients are experts in their own disease or condition and the ultimate consumers of medical products

- Patient experience data can inform:
  - Decision by sponsor to move program to clinical stage
  - Design of clinical trials
  - Selection and/or development of endpoints
  - Assessment of benefit-risk during regulatory reviews

- Anyone can collect and submit patient experience data, including
  - Patients
  - Family members and caregivers of patients
  - Patient advocacy groups
  - Disease-specific research consortiums
  - Researchers
  - Industry sponsors

PFDD Meetings: Example of FDA's Use of Patient Experience Data

The PFDD initiative started in 2012 as part of FDA's commitments under PDUFA V.

- FDA-led PFDD meetings
    - FDA recognized there are many more diseases/conditions than can be addressed in a reasonable time by FDA.
- To help expand the PFDD initiative, FDA introduced externally-led (EL-PFDD) meetings in 2015.
    - Planned and hosted by patient organizations with input of FDA staff

PFDD meetings target diseases with the following characteristics:

- Identified need for patient input
- Chronic, symptomatic, affect daily function
- Have aspects that may not be recognized or formally captured in clinical trials
- Have no/very few therapies that impact how a patient feels, functions, or survives
- Have a severe impact on identifiable subpopulations (such as children or the elderly).

PFDD Meeting Format

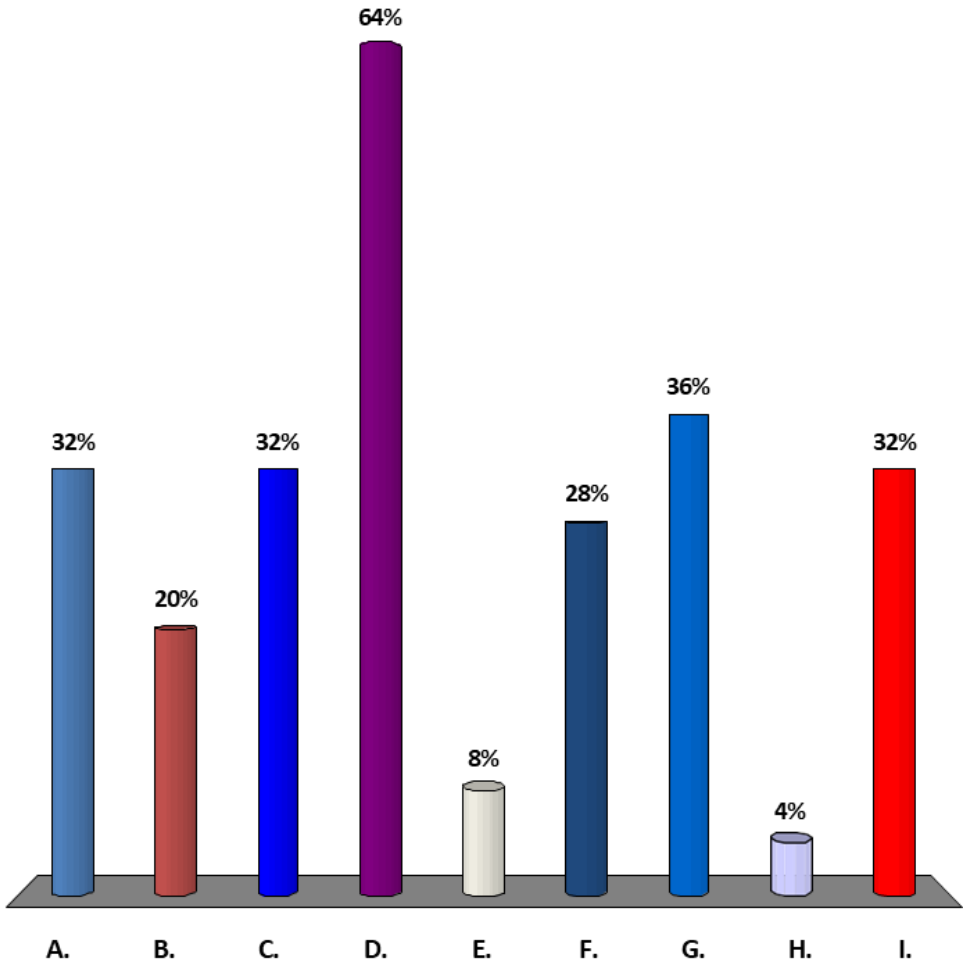
- Town hall where patients and caregivers discuss their perspectives
  - Two panels followed by open discussion
    - A few patients and caregivers share their individual stories before audience members and online participants provide input
    - Open discussion intended to provide diverse perspectives
- Panel 1 covers symptoms and impacts of the condition
- Panel 2 covers current treatment options and expectations for a new treatment, including benefit-risk assessment and clinical trial participation
- Following each PFDD meeting, FDA summarizes the discussion in a “Voice of the Patient” report that is made public
  - For EL-PFDD meetings, the patient group is responsible for summarizing

PFDD Meeting on Autism Hosted by FDA in May 2017

- Older but relevant example for condition with significant functional impacts
- Topic 1: Health Effects and Daily Impacts of Autism
  - What health effects are most challenging for you/your child?
  - How do these health effects impact your/your child's life day to day?
  - How have your/your child's experiences with autism changed over time?
- Topic 2: Current Approaches to Treatment
  - Are you/your child currently pursuing any interventions or treatments for autism? If so, what are your/your child's goals for treatment?
  - How well do your current treatments meet these goals?
  - What would you consider to be a meaningful benefit of any treatment? What type and how much improvement would be impactful?
  - What are the key things you think about when deciding whether to start a new treatment?

Which health effects of autism are most challenging to you/your loved one?

A. Irritability or disruptive behaviors
B. Cognitive impairment
C. Social Impairments
D. Communication difficulties
E. Repetitive Behaviors
F. Sleep Issues
G. Depression or Anxiety
H. Gastrointestinal symptoms
I. Other health effects not mentioned



What aspect of your/your loved one's daily life are most negatively affected by autism?

A. Ability to participate or perform daily activities (work, school, sports, drive, hobbies)

B. Ability to care for self or family

C. Risks to safety of self or others

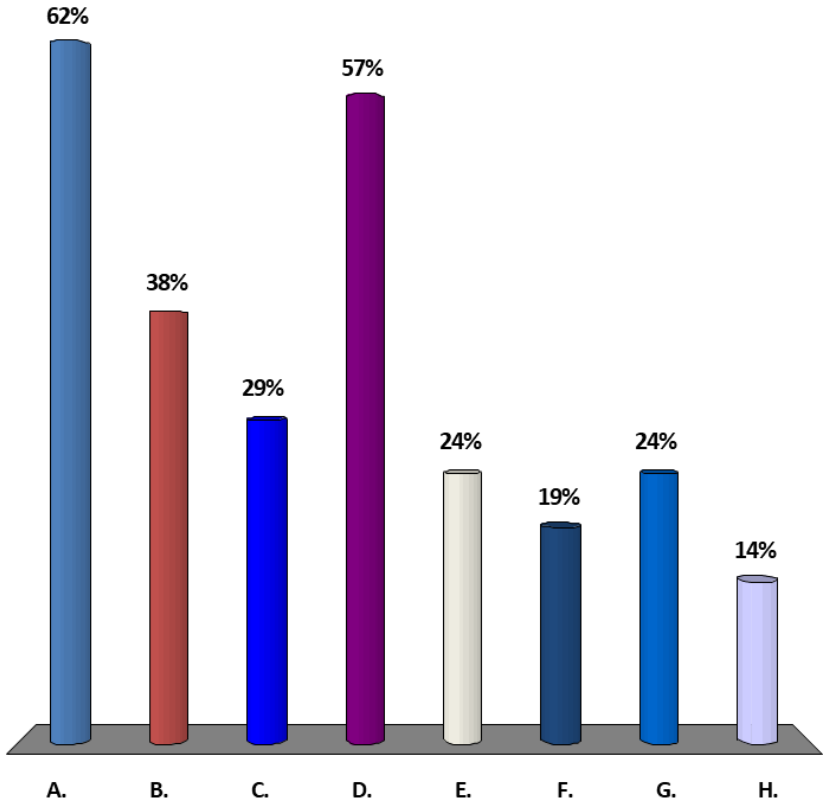
D. Impact on relationships with friends and family

E. Stigma and social discrimination

F. Emotional impacts

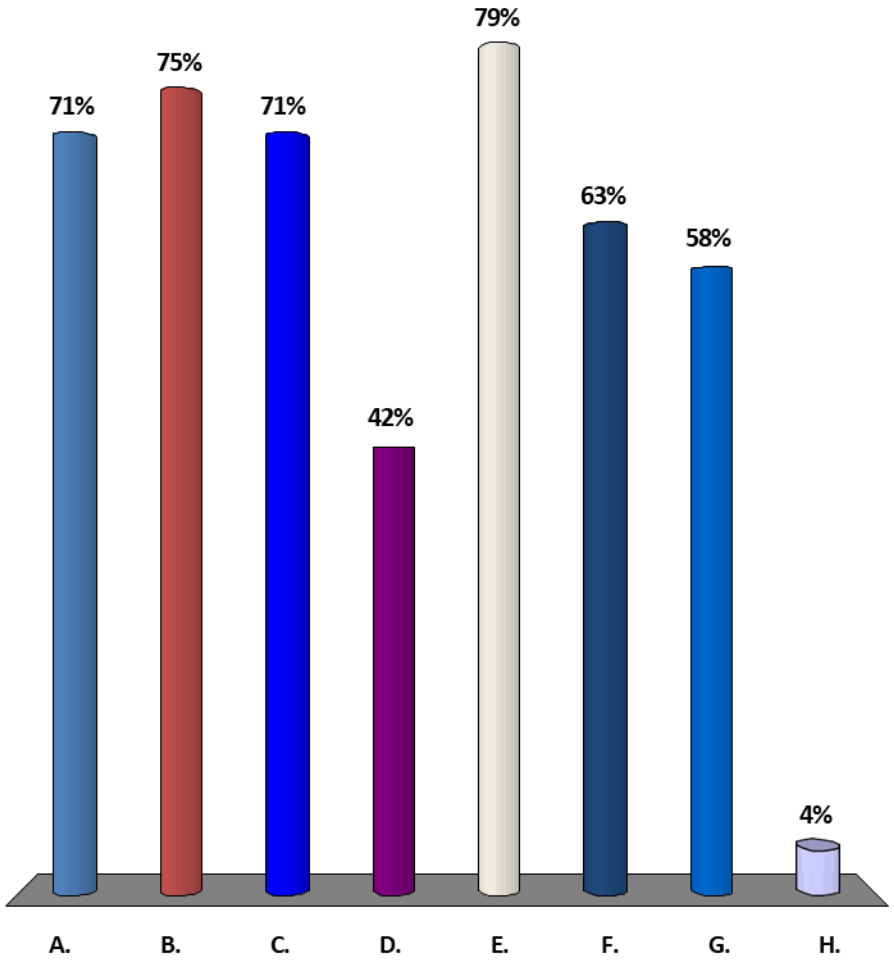
G. Burden of medical care

H. Other impacts not mentioned



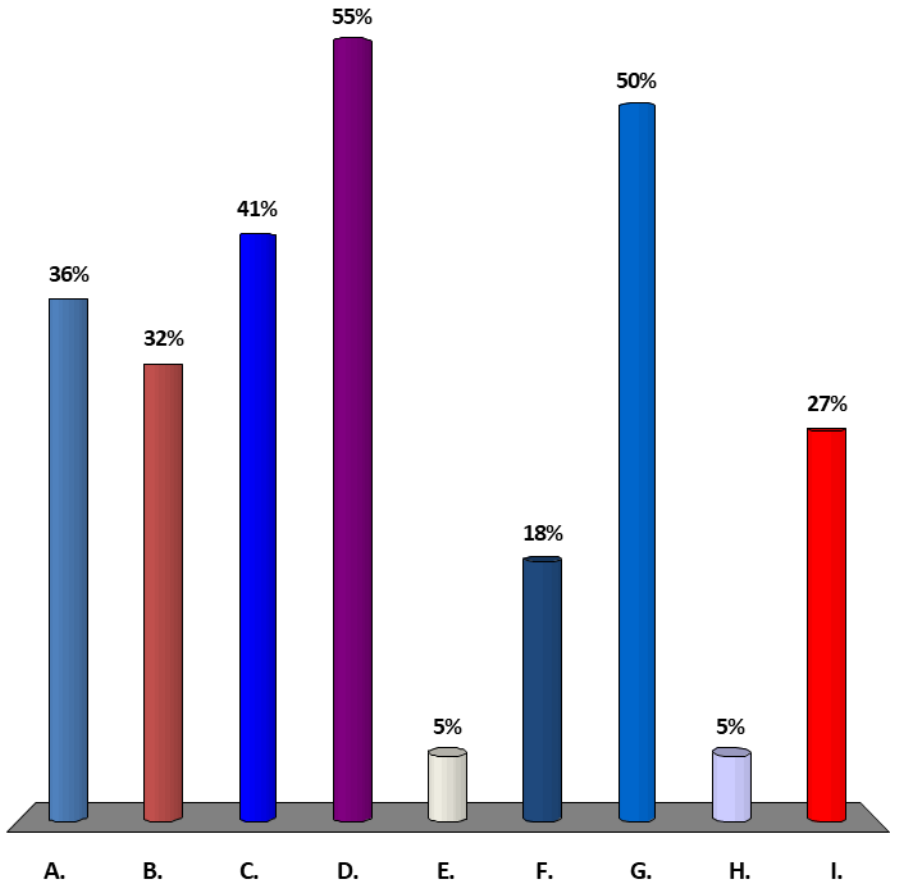
Have you/your loved one ever used any of the following to help reduce symptoms of autism?

A. Prescription medications (such as anticonvulsants or psychiatric medications)
B. Psychotherapy and behavioral therapy (such as counseling or support groups)
C. Speech therapy
D. Physical therapy
E. Occupational therapy
F. Diet modifications
G. Other therapies not mentioned
H. I've never used any therapies



When considering treatment options, which of the following benefits would you/your loved one consider to be most important?

A. Reduced irritability and/or disruptive behaviors
B. Reduced cognitive impairment
C. Reduced social impairments
D. Reduced communication difficulties
E. Reduced repetitive behaviors
F. Reduced sleep issues
G. Reduced depression or anxiety
H. Reduced gastrointestinal symptoms
I. Other



Hypothetical Scenario

Imagine that a new oral medication indicated to treat a core symptom associated with autism has recently been approved by FDA. Your doctor believes that you/your loved one may be a good candidate for this medication.

The medication requires administration every six hours and evaluation every eight weeks for dosage adjustment. The tablet can be crushed and mixed with food for ease of administration.

Common side effects of this medication include drowsiness, diarrhea, nausea and insomnia. Serious side effects such as respiratory tract infections and blood clots are rare, but possible.

What first thoughts come to mind as you hear this scenario?

What questions would you ask your doctor about this treatment?

Which of the following have the biggest impact on your decision to use a medication to help manage autism?

A. How the medication is administered
B. The frequency and length of treatment
C. Your access to treatment (such as insurance coverage)
D. Whether the medication was studied in children
E. How much the medication showed benefit for a specific symptom
F. Common side effects
G. Rare, but serious side effects
H. You/your loved one's previous response to a similar treatment
I. Whether other treatment options are available

EL-PFDD Meeting Request

- FDA requires a Letter of Intent (typically 3-4 pages) that addresses:
  - The importance of the meeting in the context of the disease area
  - Meeting goals, key areas of learning, discussion questions, patient engagement plans and proposed method for disseminating the results
  - Requests for specific FDA attendees who may attend remotely
- The LOI should be submitted approximately 1 year before the expected meeting date
- If approved, FDA recommends holding the meeting in conjunction with an annual conference or symposium to maximize attendance and get more diverse feedback
- Information contained in the LOI, including the name of your organization and a point of contact, is shared on the FDA website
- For more information on requesting a PFDD meeting patientfocused@fda.hhs.gov

Upcoming Externally-Led PFDD Meetings

Disease or Condition	Organization Submitting LOI	Organization Contact	Anticipated Meeting Date
Sarcoidosis	Foundation for Sarcoidosis Research	Tricha Shivas tricha@stopsarcoidosis.org (mailto:tricha@stopsarcoidosis.org)	October 28, 2024
Pre-symptomatic and Early to Mid-stage Adult Onset of Huntington's Disease (HD)	Huntington's Disease Society of America (HDSA)	Phyllis Foxworth pfoxworth@hdsa.org	November 13, 2024
Angelman Syndrome	**Foundation for Angelman Syndrome Therapeutics (FAST) and the Angelman Syndrome Foundation (ASF)**	**Amanda Moore and Ryan Fischer** **ASPFDD@angelman.org**	**January 29, 2025**
Autoimmune Pulmonary Alveolar Proteinosis (aPAP)	National Organization for Rare Disorders (NORD)	NORD events@rarediseases.org	February 5, 2025
Phenylketonuria (PKU)	National PKU Alliance (NPKUA)	Catherine Warren catherine@npkua.org	May 8, 2025

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Questions?

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Thank You