INTRODUCTION
Rett syndrome is a complex, progressive neurodevelopmental disorder that impacts multiple organ systems from childhood into adulthood. A recent report found that 70% of individuals with typical Rett syndrome survive to at least 50 years of age. Patient-focused drug development (PFDD) The US Food and Drug Administration (FDA) established the PFDD initiative to devote attention to gathering patient perspectives. Patient experience data are critical to the development of treatments for rare diseases by:
- Providing background and context about the disease
- Informing clinical trial design, including primary endpoints
- Supporting regulatory decision-making, especially the risk-benefit assessment of a potential new treatment

OBJECTIVE
To explore the perspective of caregivers of adults with Rett syndrome and to inform the clinical development of an investigational gene therapy.

PARTICIPANTS/METHODS
Qualitative insights were collected through a workgroup:
- N=142 caregivers of women with Rett syndrome aged ≥18 years
- Recruited in partnership with patient advocacy organizations

Quantitative insights were gathered through a survey:
- N=7 caregivers of women with Rett syndrome aged ≥18 years
- 22 multiple choice and open response questions

Qualitative insights were collected through a workgroup:
- N=7 caregivers of women with Rett syndrome aged ≥18 years
- Recruited in partnership with patient advocacy organizations
- Daily online survey, including journaling exercises, asynchronous discussion forum; video focus group

RESULTS
THE AGING CAREGIVER EXPERIENCE
As caregivers age, they face unique challenges with mobility and concerns about planning for their loved one’s future.

PHYSICAL
- Progressive mobility issues in adulthood increase caregiver burden
- Reports of back pain, fatigue, and falls (for both) were common

EMOTIONAL
- As the disease progresses, continuing therapy becomes challenging, leaving caregivers to wonder “what can I do?”
- Behavioral or mood disturbances causes caregivers to question attending social activities or to isolate the family

FINANCIAL
- Caregivers’ biggest fear: “if something happens” to them who will care for their loved one
- Access to high-quality, affordable day programs for adults is limited

CHALLENGING SYMPTOMS THAT AFFECT QUALITY OF LIFE
Caregiver-rated symptoms with greatest impact on quality of life are highlighted. The top 5 are listed below in order, starting with the most challenging:

- Loss of speech
- Involuntary hand movements
- Sleep disturbances
- Hyperventilation and breath holding
- Hypertonia

Note: Green boxes represent those treatments or interventions used by 40% or more of respondents in each respective age group

“The who will care for her? Where will she live? Do we have enough money saved for her care?”

“Youngest fear is not being able to take care of her. I worry something happens to me or my husband.”

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Accessed on 3/14/22

Open 2020;4:e000717. doi:10.1136/bmjpo

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