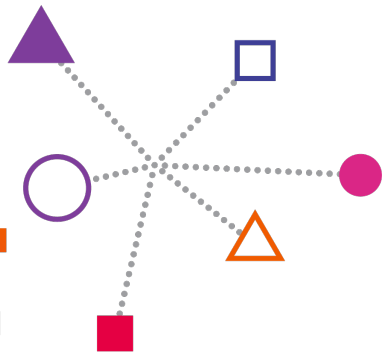


EAF

European Achondroplasia Forum



Advances in Achondroplasia 2023

21–22 April 2023, Frankfurt



MEASURING OUTCOMES IN ACHONDROPLASIA

Valérie Cormier-Daire and Marco Sessa

Disclosures

- Marco Sessa: BioMarin
- Valerie Cormier-Daire:
 - Outside of this meeting, have received honoraria from BioMarin
 - Involvement in QED clinical trials



OUTCOMES OF IMPORTANCE IN INFANCY AND EARLY CHILDHOOD: EAF SURVEY DATA

Valérie Cormier-Daire



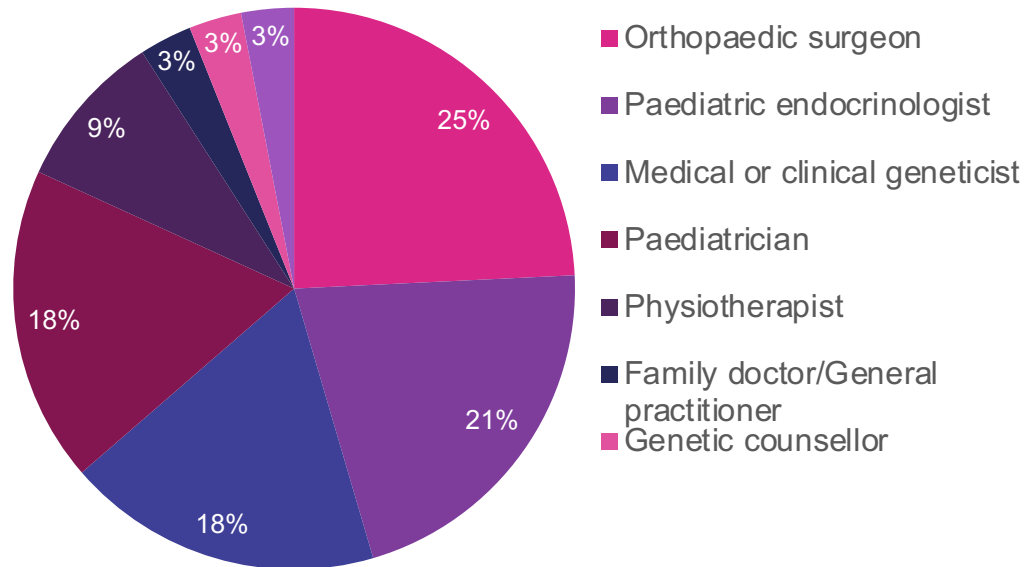
EAF Outcomes Survey 2022

- In preparation for an EAF workshop on *Clinical Challenges in Infants and Early Childhood* held in October 2022, two surveys were carried out
 - One for healthcare professionals
 - One for parents of children with achondroplasia, available in six languages
- The aim was to identify differences between HCP and parent perspectives of care
- Both surveys addressed questions in relation to infants (aged ≤ 2 years) and young children (aged 2–5 years)
- Key points addressed were:
 - Outcomes of importance
 - Clinical challenges
 - Awareness of and adherence to guidelines and recommendations for care (HCPs only)

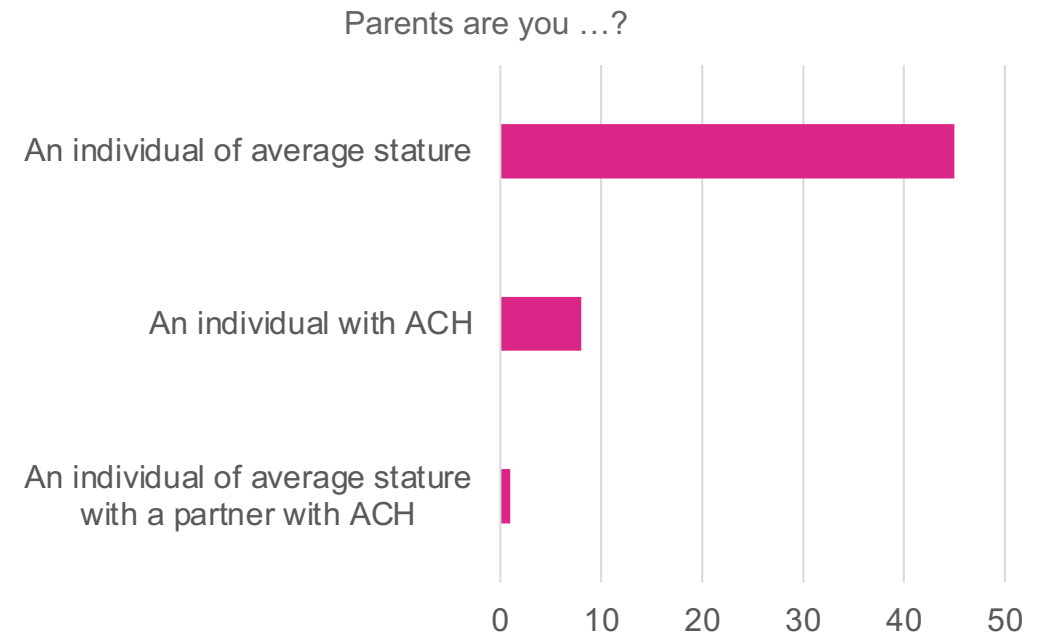


Respondents

- 33 HCP respondents from 22 countries
- 75% based in academic institutions*



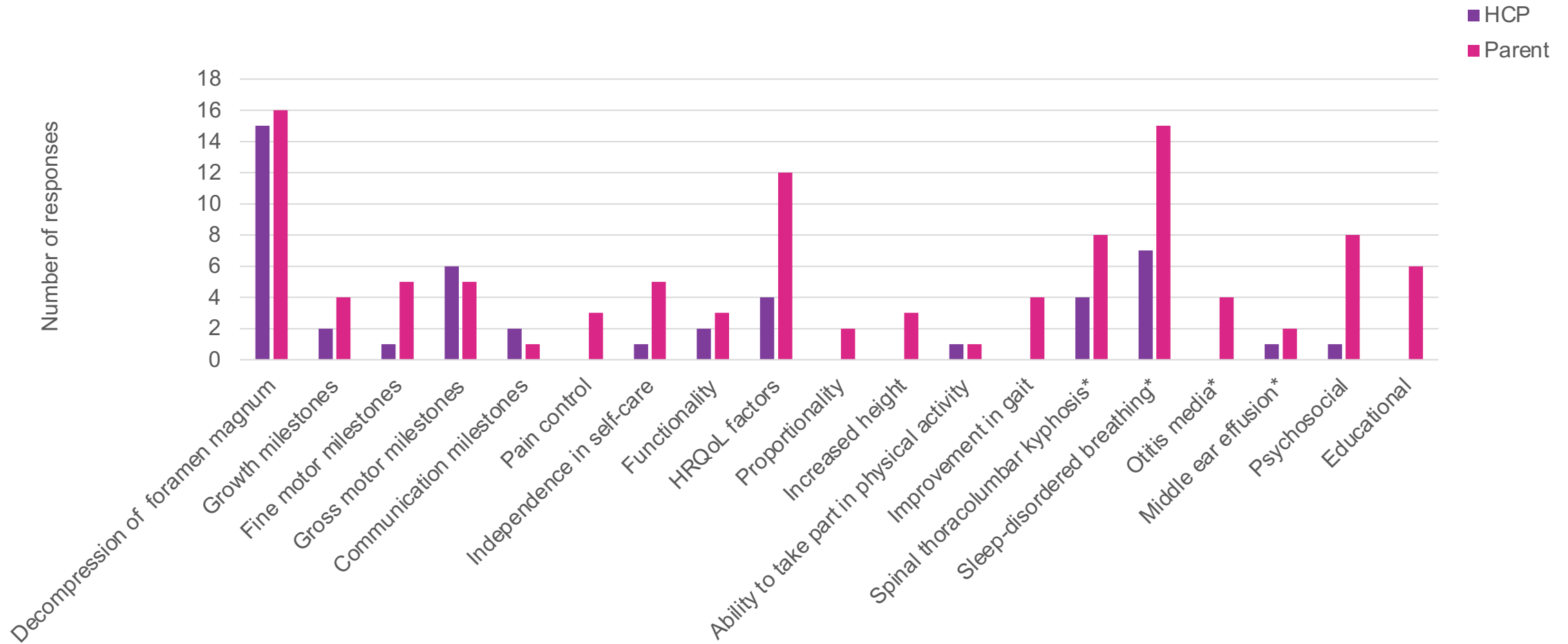
- 55 parent respondents from 13 countries
- 63% whose children receive care in academic institutions*



*Academic institution/Specialist achondroplasia centre/University hospital



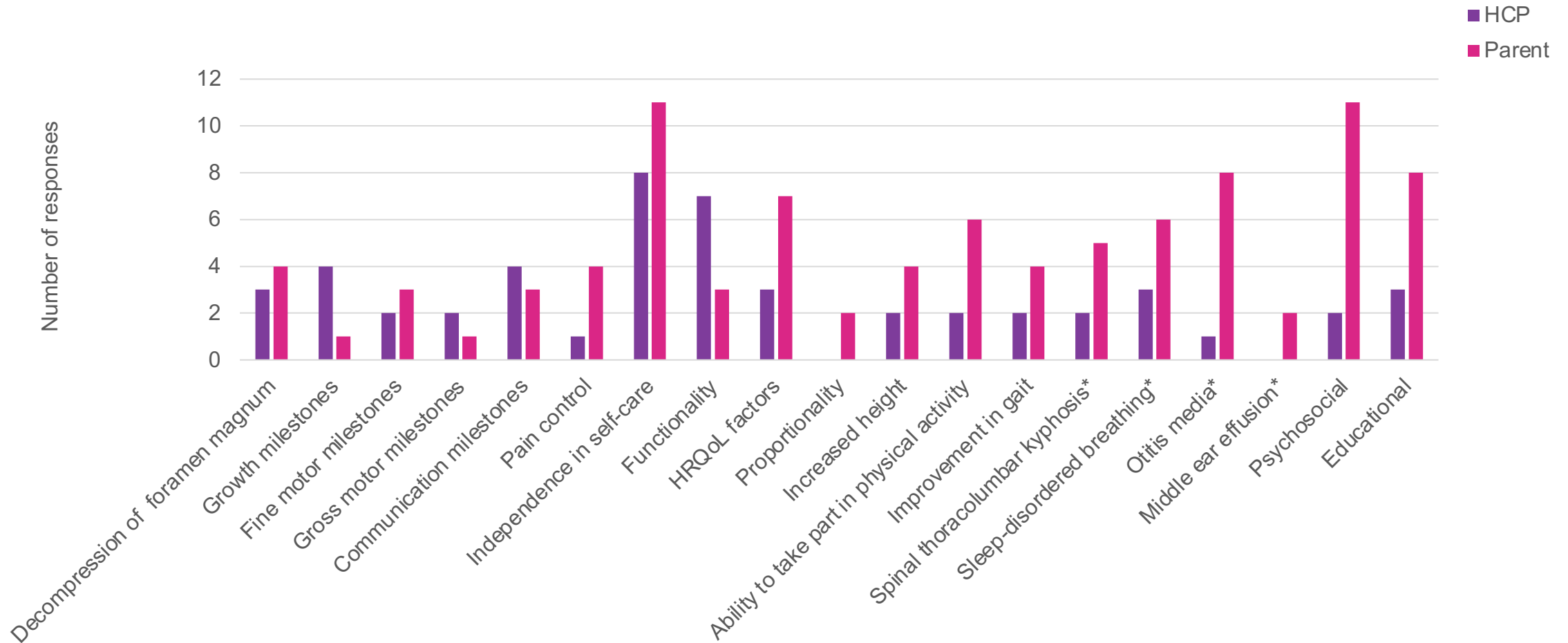
Most Important Outcomes in Infancy



*Resolution of ...; All milestones are ACH-specific; Psychosocial (acceptance and understanding of condition); Educational (supporting provision of appropriate support in school (physical, psychological, educational))



Most Important Outcomes in Early Childhood



*Resolution of ...; All milestones are ACH-specific; Psychosocial (acceptance and understanding of condition); Educational (supporting provision of appropriate support in school (physical, psychological, educational))



EAF Survey Conclusions

- HCP's priority is to address medical interventions, particularly in infancy when complications can be life-threatening
- Parent's priority is more focused on QoL, especially in later childhood, once threats to life are diminished



EXPECTATIONS OF TREATMENT

Valérie Cormier-Daire



New Therapies Lead to New Expectations

- Current treatment options for ACH are known and understood
- All have a target outcome, with some additional benefits
 - E.g. Surgical interventions to address deformity, leading to improved function
- The advent of vosoritide has led to the expectation of benefits beyond its licensed outcome of augmenting linear growth
 - Expected additional benefits may lead to a perceived improvement in QoL
- Novel treatments in development may lead to further expectations



Key Questions to Discuss Today

- What are the expectations of treatments?
 - HCP perspective
 - Patient/parent perspective
- Are these realistic?
- How do those expectations translate into QoL improvements?
 - HCP perspective?
 - Patient/parent perspective



FROM CURING YOURSELF TO CARING FOR YOURSELF: QOL OF PEOPLE WITH ACH

Marco Sessa



ASSUMPTIONS:



- ACH is:
 - A rare bone disease caused by a genetic mutation in the fibroblast growth factor receptor 3
 - **Lifelong**; considered a chronic condition
- **Disease state is not continuous**; reduces to few or no episodes over the lifespan in many people living with achondroplasia
- What is particularly relevant is **physical appearance and functional diversity**
 - Can cause psychosocial-emotional distress for many children, adolescents, and adults with ACH, as well as their families ...
- The great difficulty is **stigma** more than height
 - Others may associate people with achondroplasia with fantasy characters (dwarves), rather than see them as a person
 - This cultural heritage is very heavy for us; it demands a lot of energy, effort, and stress



Criteria to measure QoL of people with ACH changes depending on subject involved



- For healthcare system: Well-being
 - ▲ Physical autonomy
 - Height to be able to ring a doorbell
 - Ability to pick up an object

- For parents: Cure
 - ▲ Perception of short stature on their children
 - Difficulties building friendships
 - Lower self-esteem

- For people with ACH: Care
 - ▲ Social autonomy
 - Don't need 'curing'
 - To not be seen as a patient/diseased





What Does QoL mean for Health Care Systems?

- To achieve a good level of health and well-being through cure and attention
 - Reaching autonomy parameter: height and *self-sufficiency* (i.e. for personal hygiene)
 - Frequent and constant follow up





What Does QoL mean for Parents?

- Perception of short stature on their children as the main obstacle to:
 - Having equal rights to access social life and have an autonomous life
 - Building friendships
 - Self-esteem



What Does QoL mean for People with ACH?



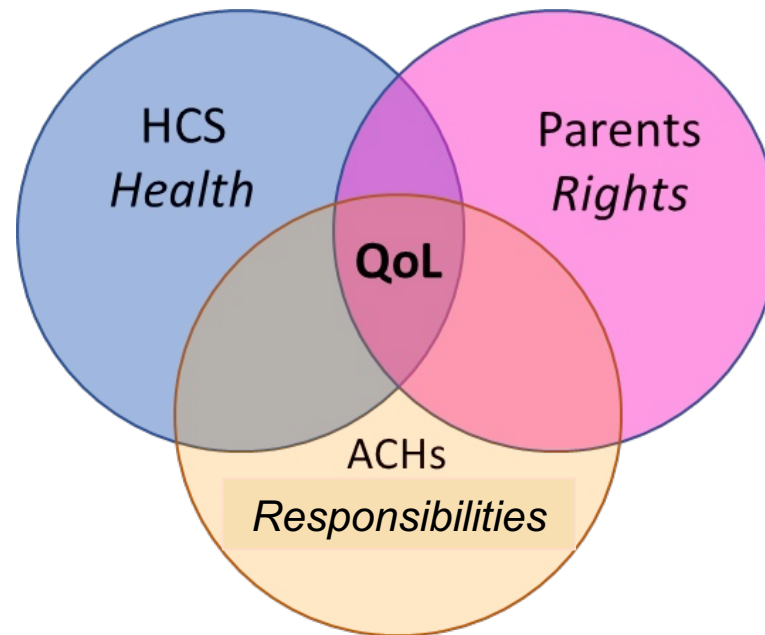
- Social autonomy
 - To not be perceived as a life-long patient/diseased
 - Reach life independence
 - Build own personality/individuality





What can we do to Improve QoL for People with ACH?

- The QoL for people with ACH is a mix of Health, Rights, and Responsibilities
- Every one of us can contribute to improve QoL in respect to the other elements



DISCUSSION



Discussion Session

- Split into groups
- Discuss key questions (10 mins on each question) to address:

What are the outcomes of importance in ACH;
how can we facilitate and measure real change in QoL?



Key Questions

Expectations of treatment

- What are the expectations of treatments?
 - Patient/parent perspective
 - HCP perspective
- Are these realistic?
- How do those expectations translate into QoL improvements?
 - Patient/parent perspective
 - HCP perspective

Measuring QoL

- What tools are used to measure changes in QoL?
 - Do they measure the right thing?
 - Are they effective in capturing real changes in QoL?
- What should be measured to capture real change in QoL?



SUMMARY



Summary

What are the outcomes of importance in ACH;
how can we facilitate and measure real change in QoL?



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