

# The patient perspective on treatment for Skeletal Dysplasias

Sérgio B. Sousa and Alice Mirante

MMRC-ACH-00XXX. November 2023. The Achondroplasia Preceptorship is sponsored by BioMarin.

This is a non-promotional meeting for healthcare professionals only. This material does not have any promotional intention, and only aims to provide scientific information relating to diseases and/or healthcare. Currently, there is no product approved for use in achondroplasia in MEA/Turkey.

← → ↻ https://www.youtube.com/watch?v=JblSfoypJV4

YouTube PT Search



**Born Small**

Dwarfism and the drug that could change everything | Full episode | SBS Dateline

SBS Dateline ✓ 670K subscribers Subscribe

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## SPORT

### Ellie Simmonds: A World Without Dwarfism? - documentary explores views on new drug

By Ciaran Varley  
BBC Sport

🕒 4 April 2022 | [Swimming](#)



Ellie Simmonds has been on a journey to explore a controversial new drug for children with achondroplasia - a form of dwarfism.

"I'm worried that this drug could be the end of dwarfism altogether," says Ellie Simmonds in new BBC documentary, *Ellie Simmonds: A World Without Dwarfism?*

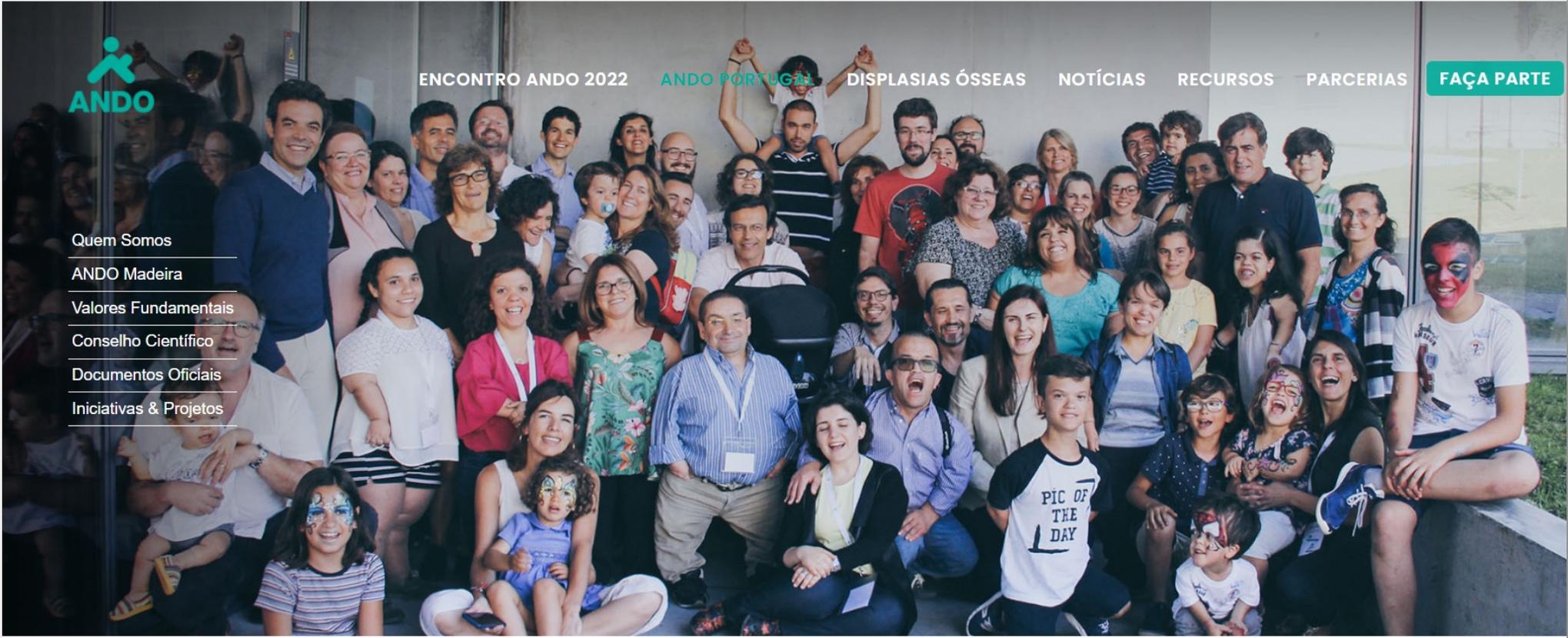
## LPA's Position Statement Regarding Biomarin's Voxzogo

Little People of America (LPA), the world's oldest and largest support organization for people with dwarfism and their families, has been tracking biotechnical developments to treat individuals with Achondroplasia. With the FDA approval of Biomarin's Voxzogo therapy, the **dwarfism community is challenged by the impact of this new treatment on our health, culture, and our standing within society.**

LPA strongly believes that a pharmaceutical intervention focusing on growth velocity is not addressing the actual health and quality of life issues of people with dwarfism. Our height-related challenges are primarily based on inaccessible architecture, lack of universal design, and society's intolerance and discrimination of people with short stature. We are concerned that this recently approved drug attempts a pharmaceutical solution to a societal issue. Our community acknowledges that the hardships associated with having dwarfism need to be addressed. Yet, we strive for our height to be reframed as a part of the diversity of humanity. We want to reprioritize research goals to be the most meaningful ones for our members, such as reducing spinal stenosis, sleep apnea, and the need for corrective surgeries.

Our role is to provide social support and advocacy for our members and their community. As part of this role, LPA supports medical research, especially that which improves the quality of life of our members by treating symptoms that can range from uncomfortable to lethal. We respect the personal choices of all families or individuals regarding healthcare decisions and welcome all individuals and families to be part of LPA, regardless of medical decisions and outcomes. As medical science moves forward, we will continue to inform researchers about the value of dwarf pride and its contributions to human biological, social, and cultural diversity.

# Crucial the dialogue with local patient associations, information given on meetings, webinars and available online



The screenshot shows the website for ANDO Portugal. The main image is a large group photo of people, including children and adults, many with face paint, suggesting a social or community event. The website layout includes a navigation menu with the following items: ENCONTRO ANDO 2022, ANDO PORTUGAL, DISPLASIAS ÓSEAS, NOTÍCIAS, RECURSOS, PARCERIAS, and FAÇA PARTE. A sidebar on the left contains links for Quem Somos, ANDO Madeira, Valores Fundamentais, Conselho Científico, Documentos Oficiais, and Iniciativas & Projetos. The footer of the page displays the ANDO Portugal logo and the breadcrumb path: > Home > ANDO Portugal.

andoportugal.org/ando-portugal/

ANDO

ENCONTRO ANDO 2022 ANDO PORTUGAL DISPLASIAS ÓSEAS NOTÍCIAS RECURSOS PARCERIAS FAÇA PARTE

Quem Somos  
ANDO Madeira  
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# Questionnaire CHUC-ANDO Aug/SepP 2023

← → ↻ https://www.andoportugal.org/displaisas\_osseas/questionarios-ando/experiencias-de-pais-e-c

ANDO PORTUGAL DISPLASIAS ÓSSEAS NOTÍCIAS RECURSOS PARCERIAS **FAÇA PARTE**

**Questionário**  
**EXPERIÊNCIAS DE PAIS E DE CRIANÇAS  
EM TRATAMENTO COM MEDICAMENTOS  
INOVADORES PARA DISPLASIAS ÓSSEAS**

**PARTICIPE!**

ANDO CHUC

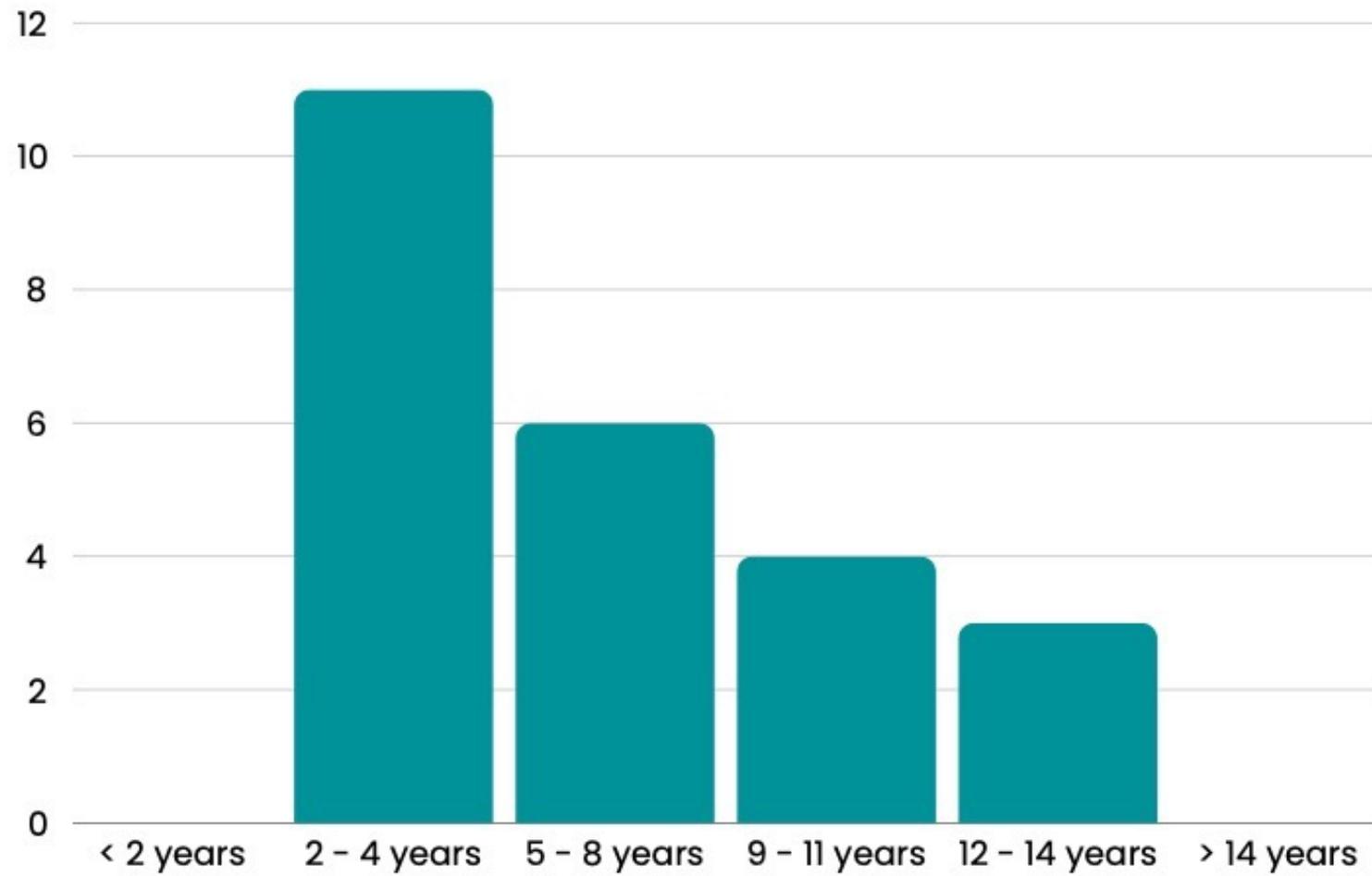
## Experiências de Pais e Crianças com Medicamentos...

> [HomeDisplasias Ósseas](#) > [Questionários ANDO](#) > Experiências de Pais e Crianças com Medicamentos Inovadores

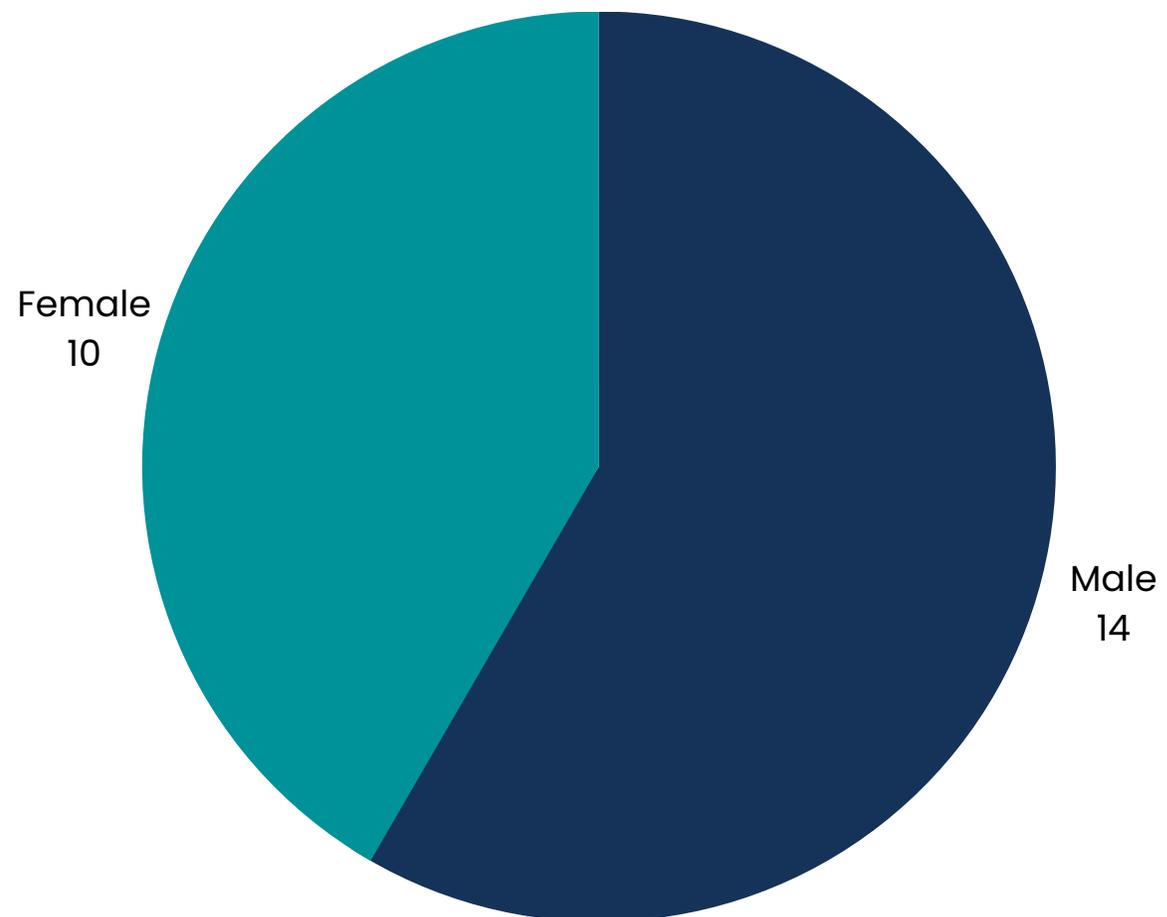
A **ANDO**, em colaboração com a **Equipa Multidisciplinar de Displasias ósseas** do Centro Hospitalar Universitário de Coimbra, elaborou este questionário para conhecer as experiências de pais e crianças no que diz respeito ao tratamento com medicamentos inovadores para as

- Anonymous
- 32 replies
- 27 children with achondroplasia
- 24 on treatment

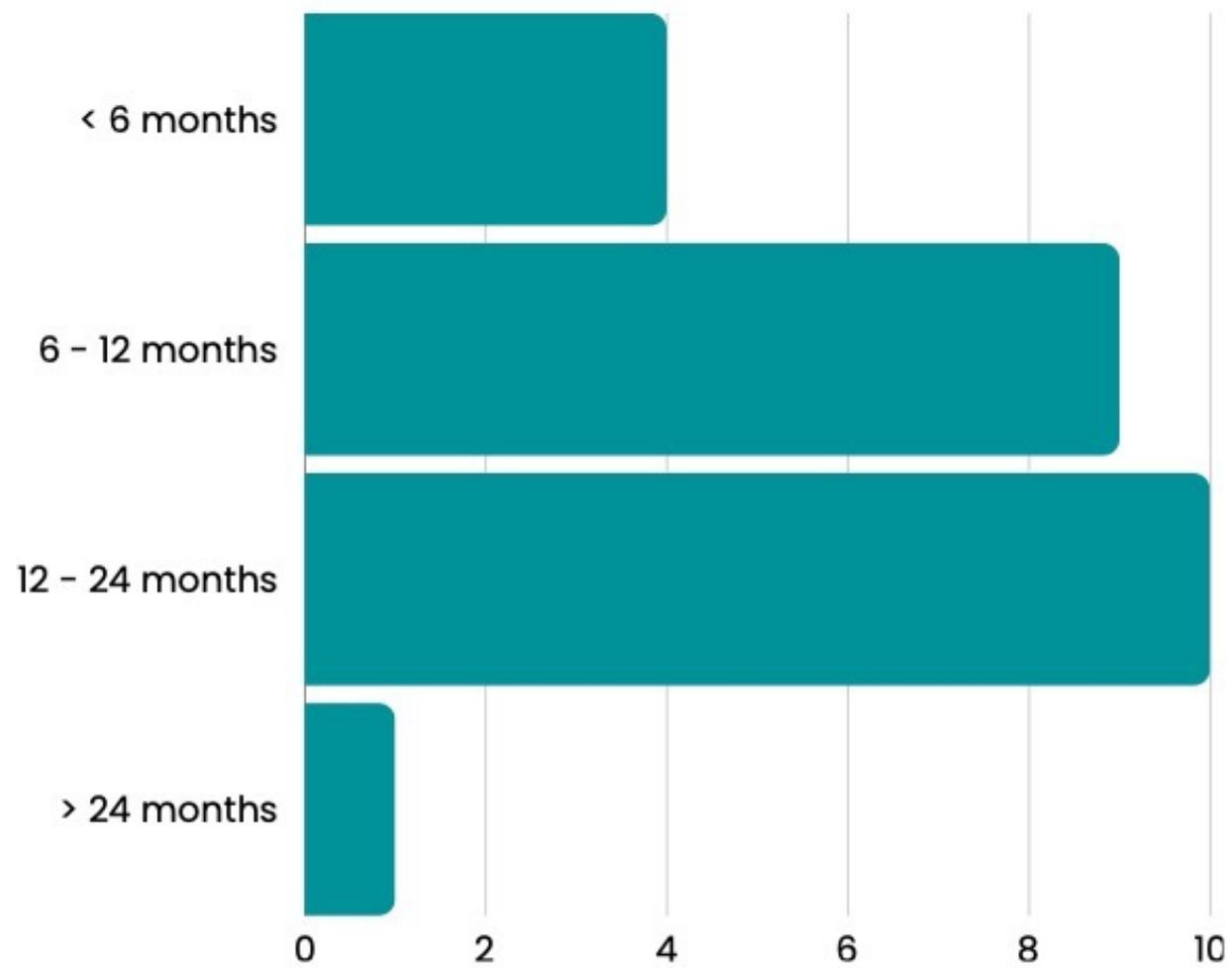
### 1. Age group of the child/adolescent



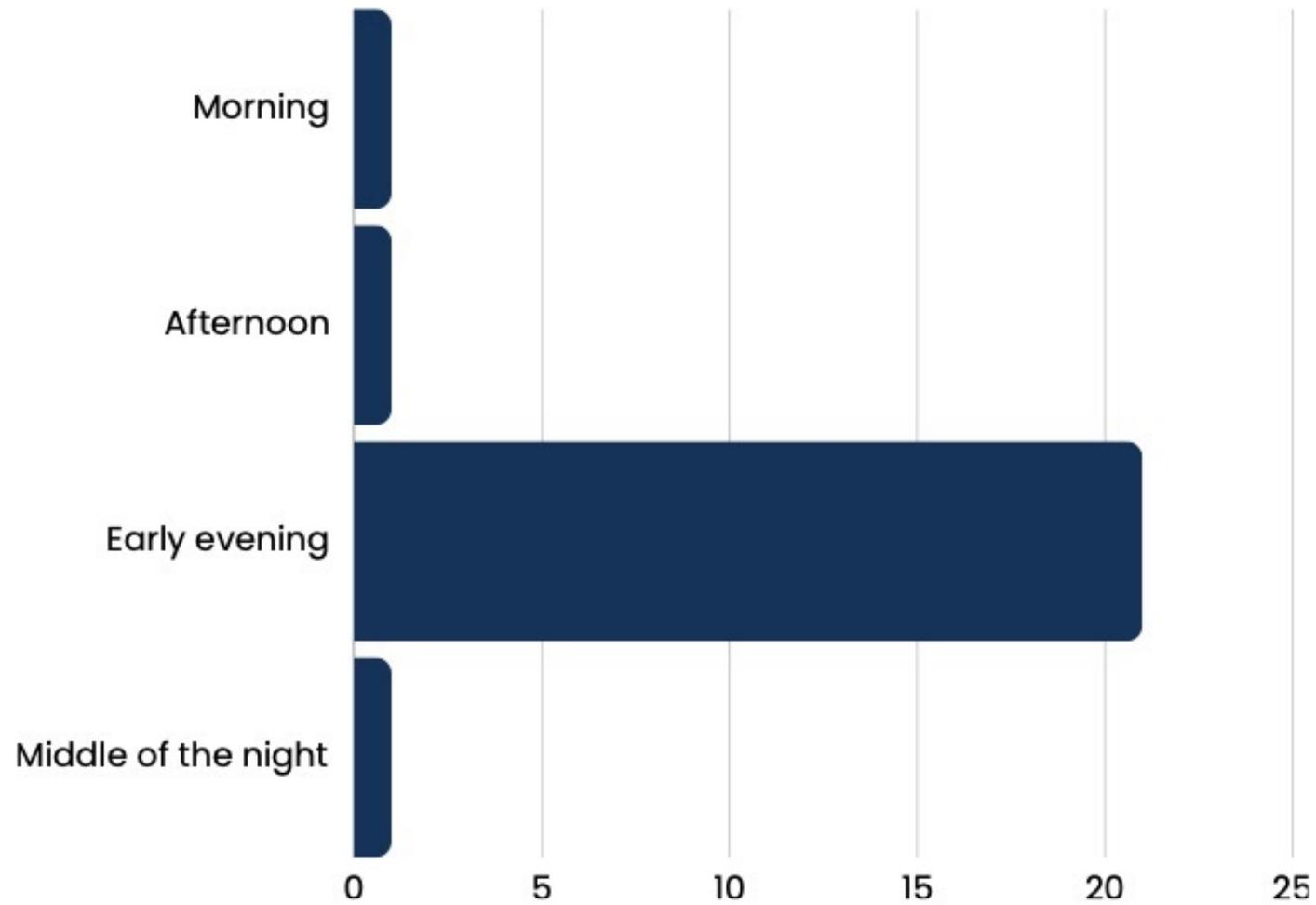
## 2. Sex of the child/adolescent



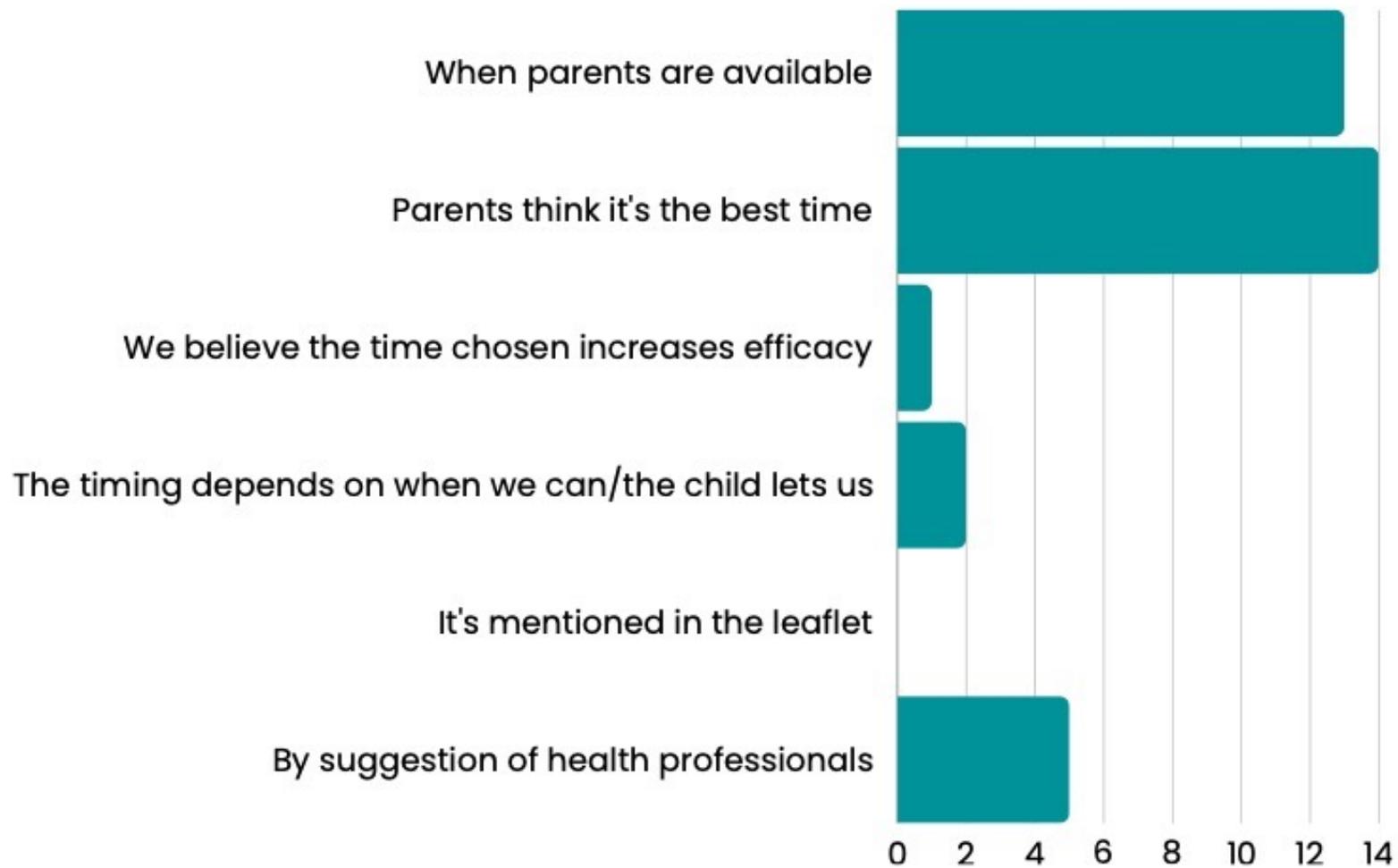
### 3. For how many months has the child/adolescent started their current treatment?



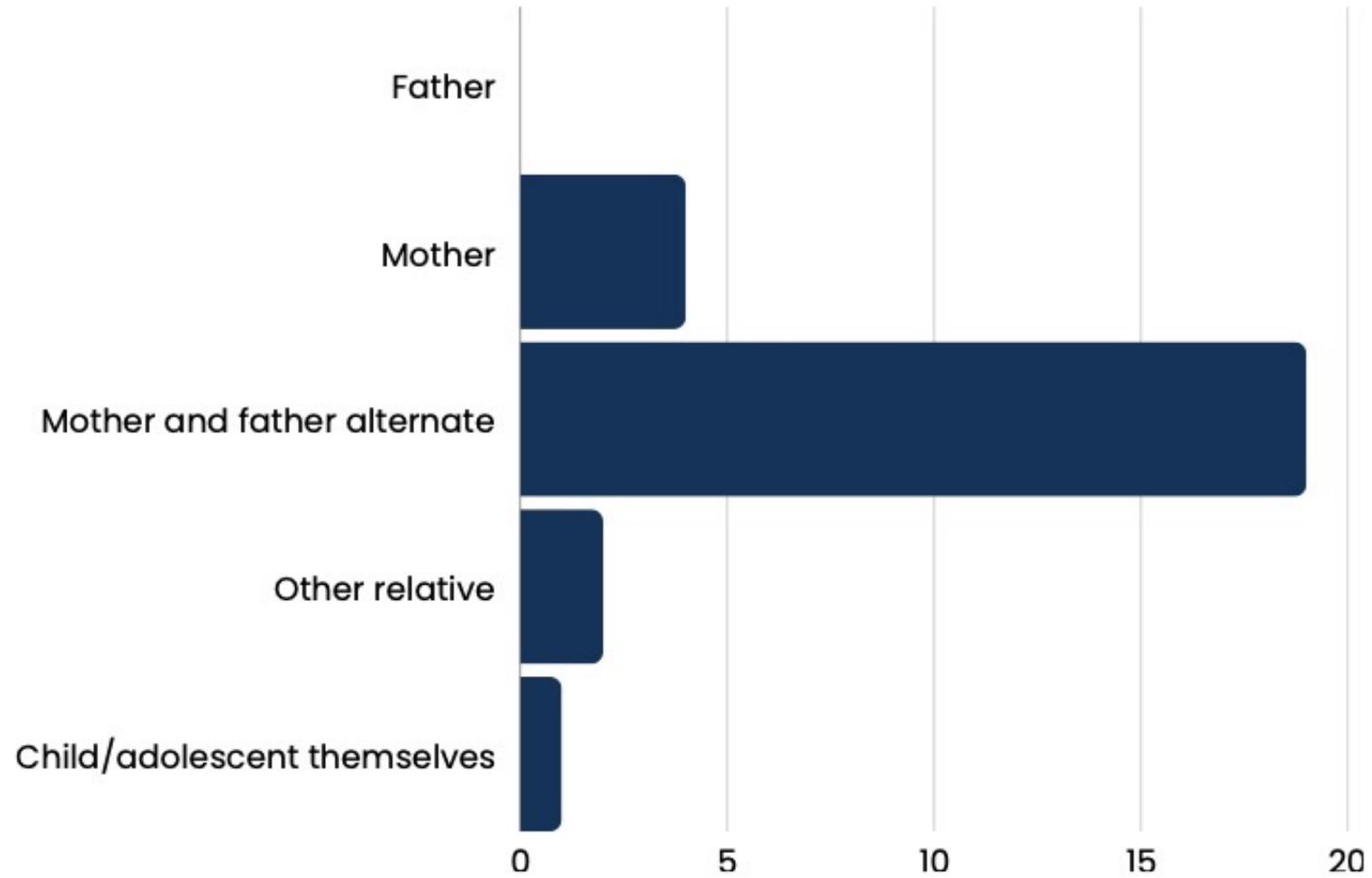
#### 4. At what time of day/night does the child/adolescent take the medicine?



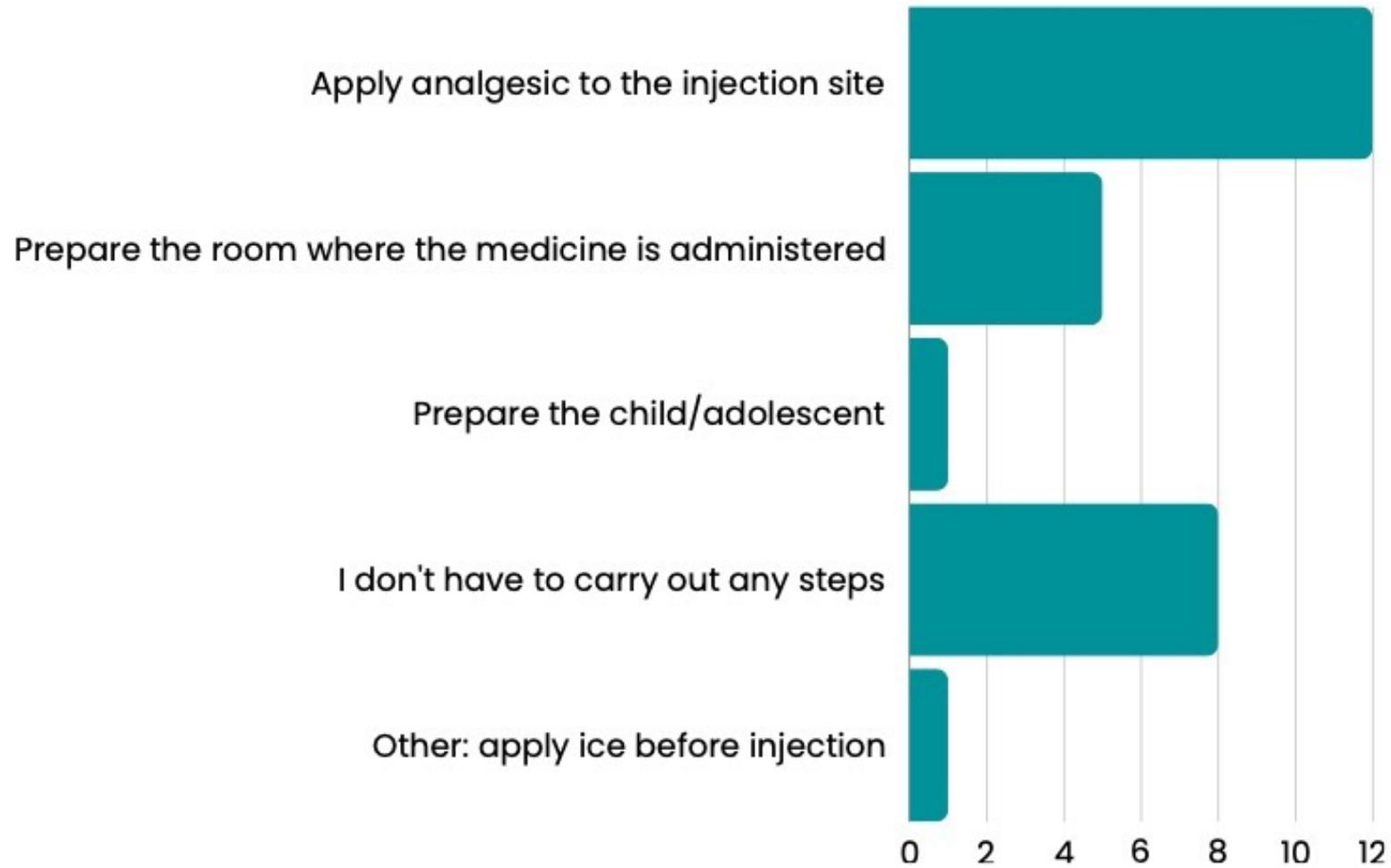
## 5. What factors influence the timing of taking the medicine?



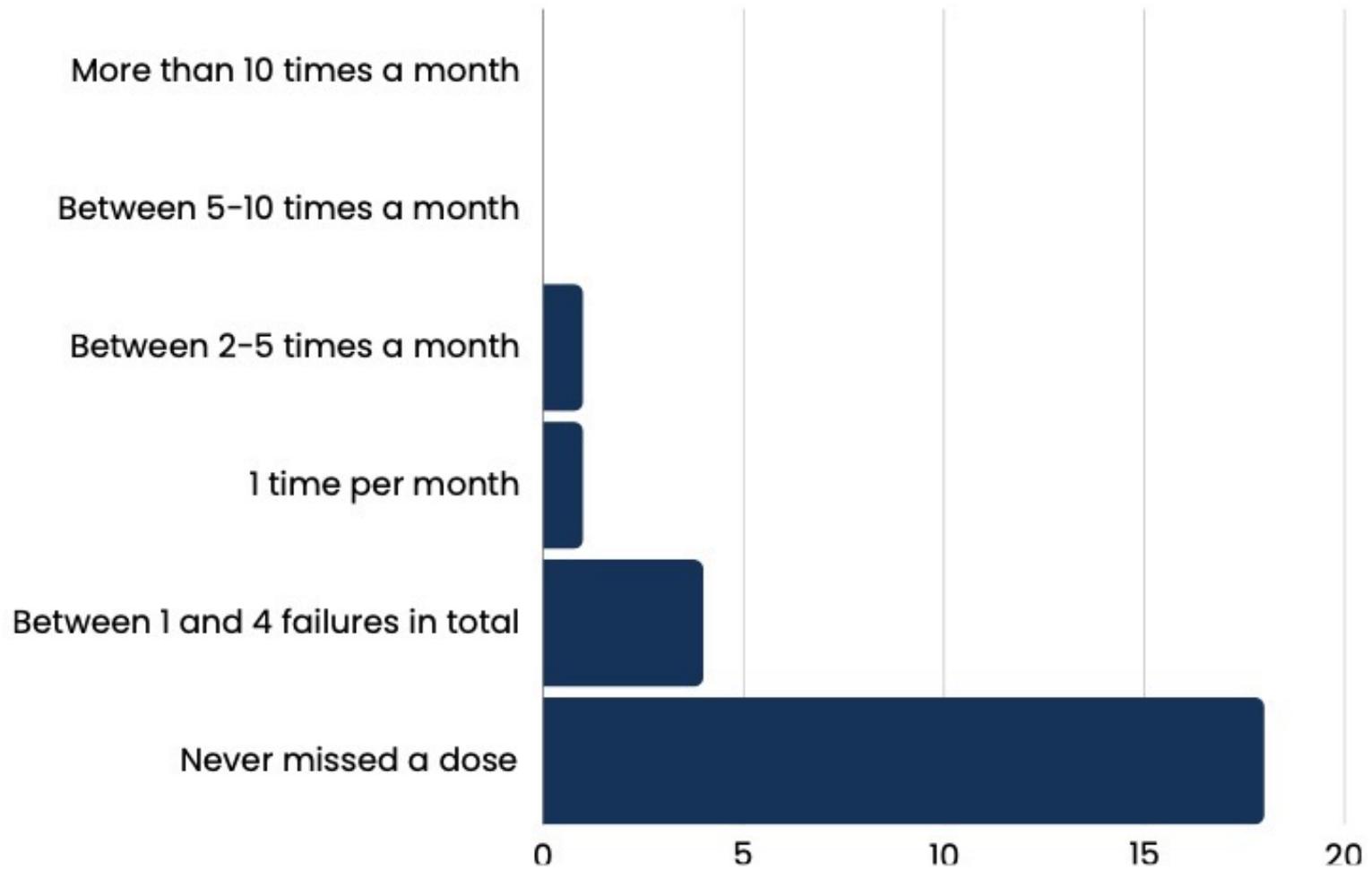
## 6. Who administers the medicine?



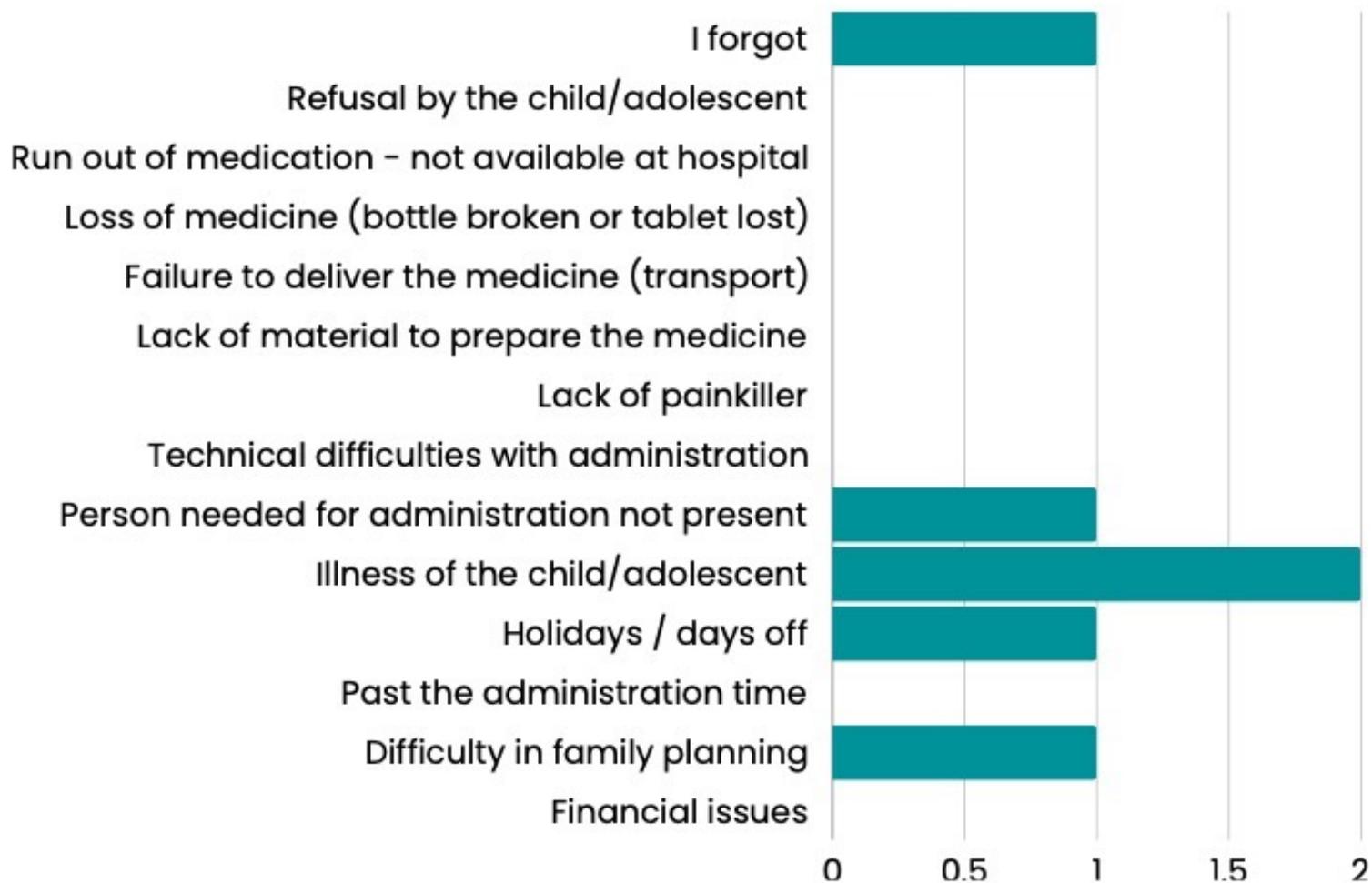
**7. Do you have to carry out any steps other than preparing the medicine?**



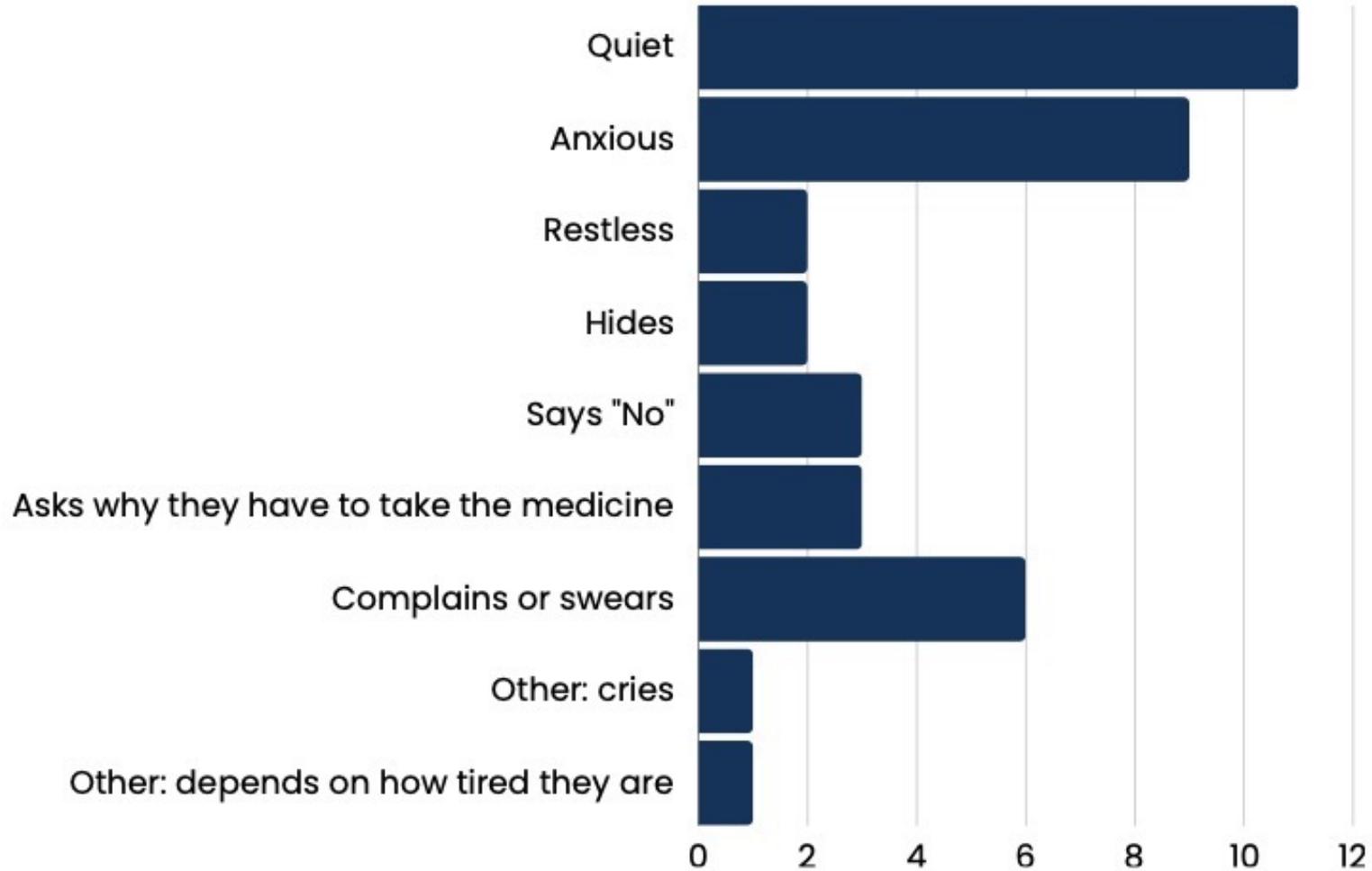
**8. Since starting treatment, state how many times you have missed a dose**



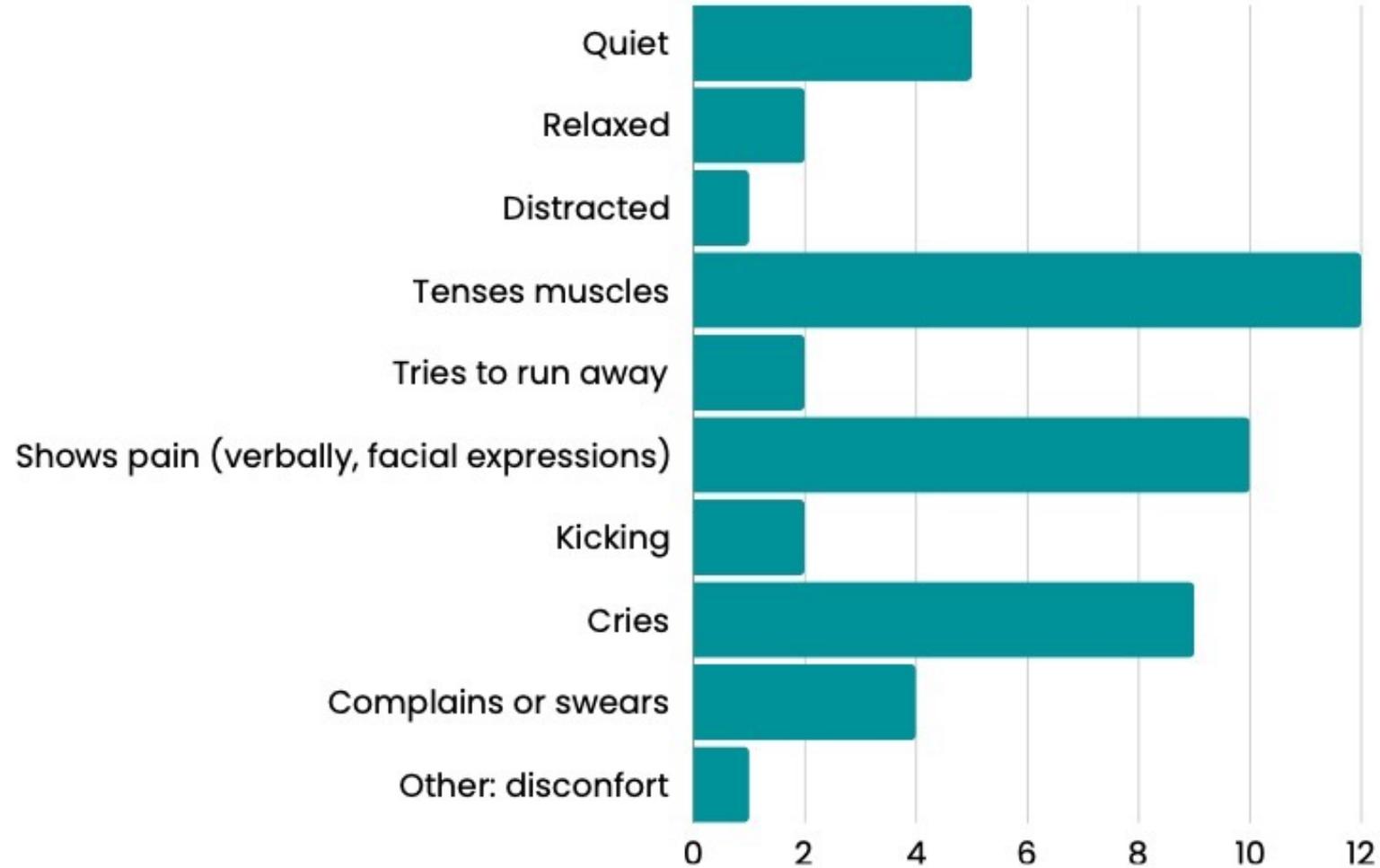
### 9. Why did you miss the dose(s)?



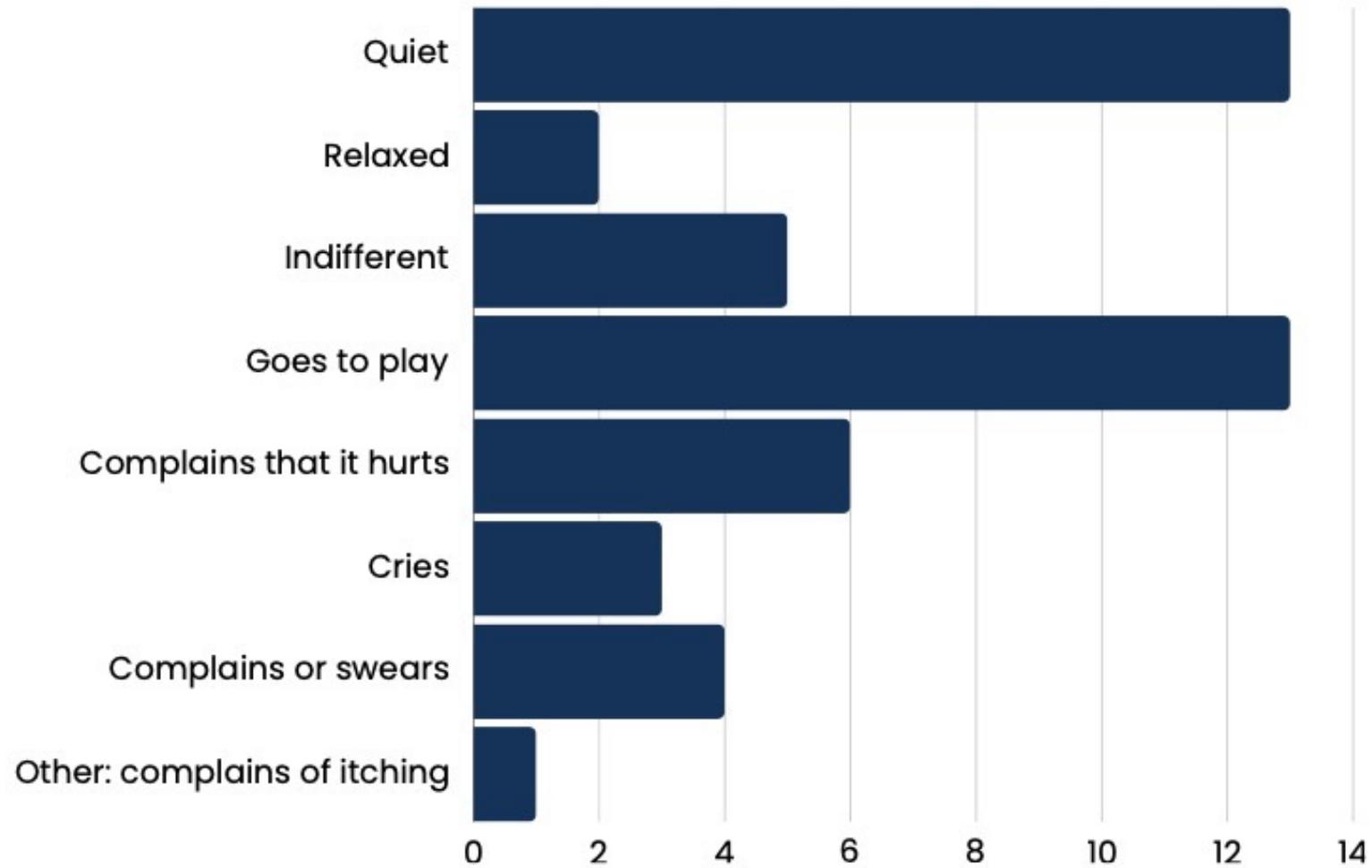
**10. Describe what you notice about the child/adolescent before the medicine is administered?**



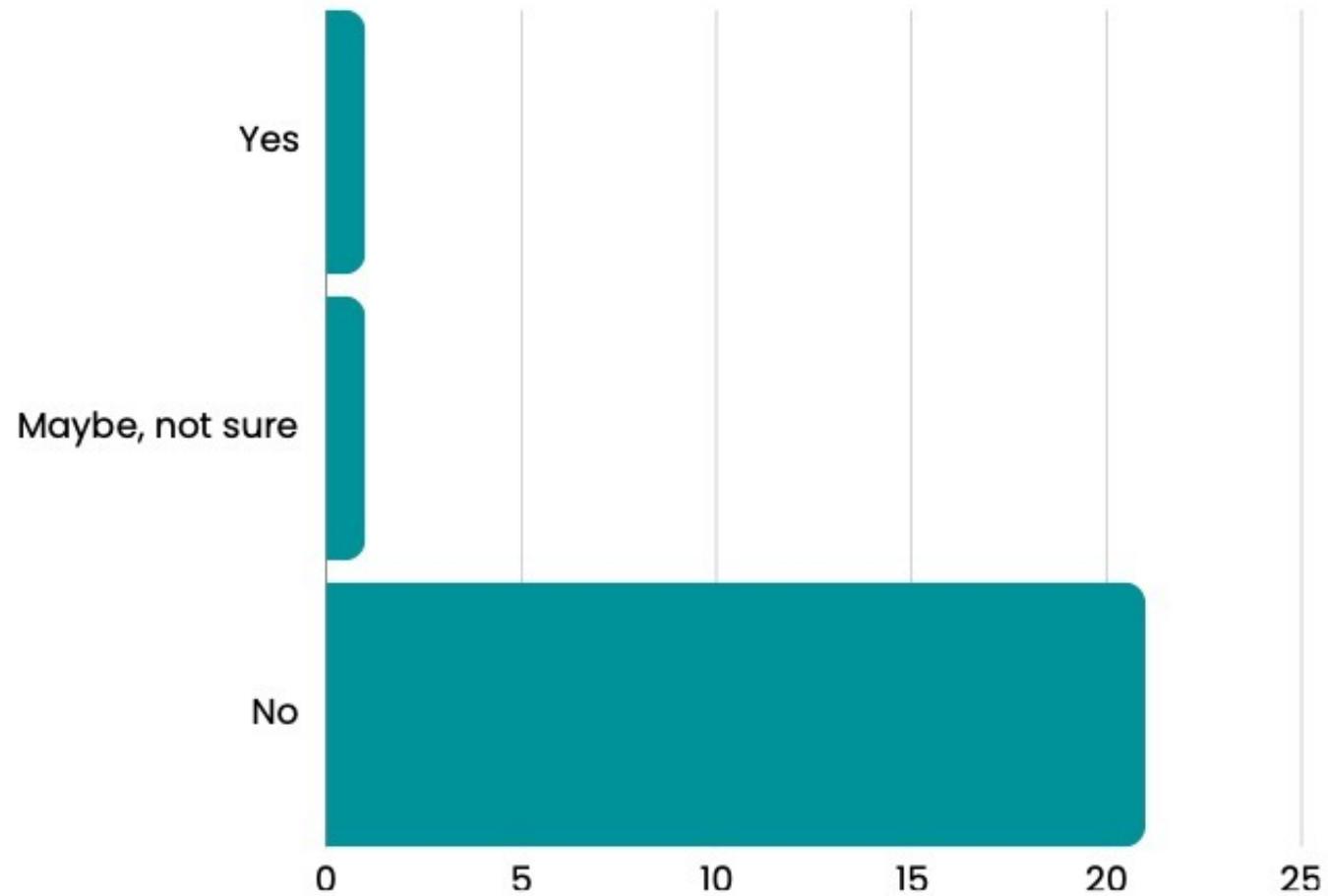
**11. Describe what you notice about the child/adolescent during the administration of the medicine?**



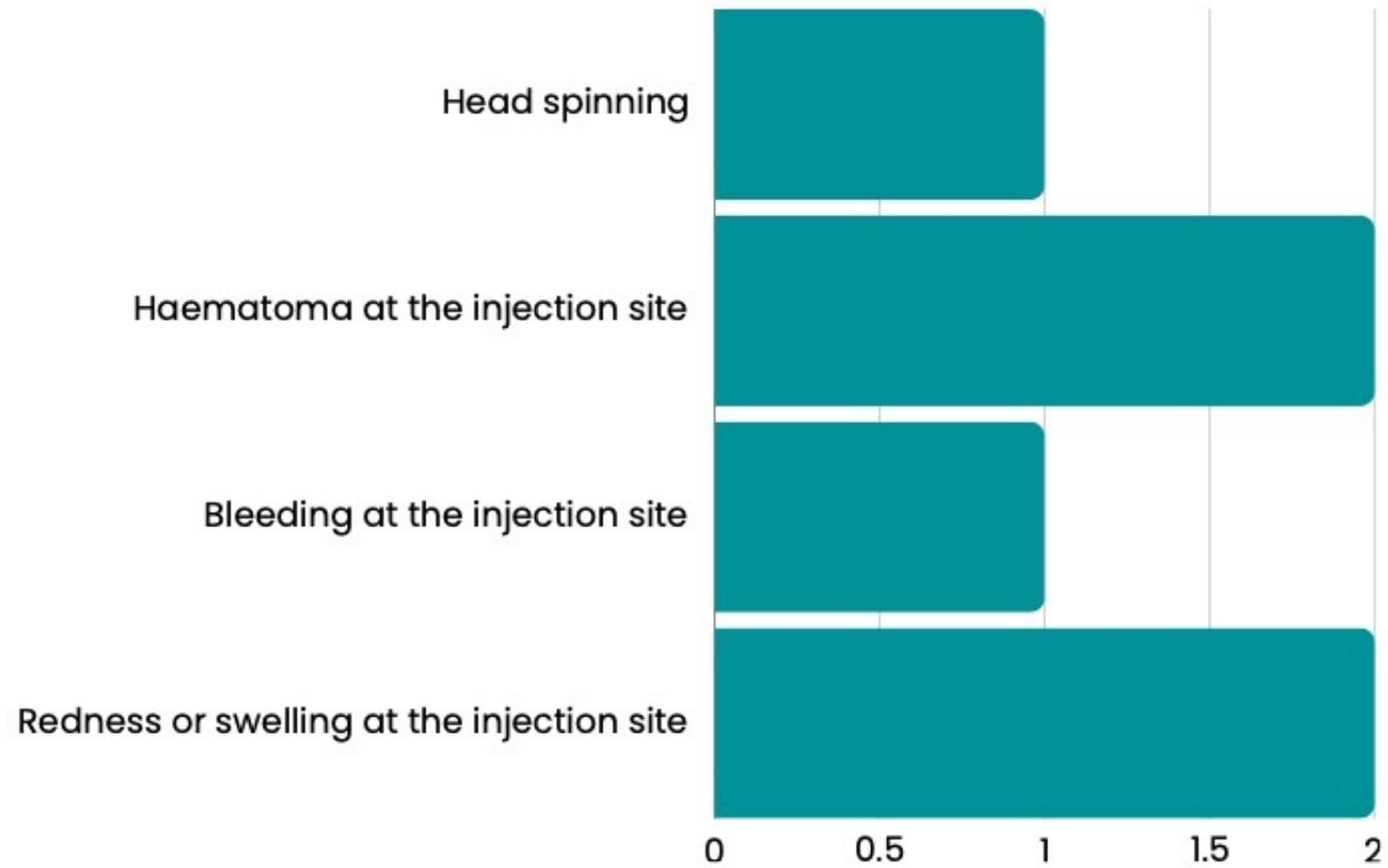
**12. Describe what you notice about the child/adolescent after the administration of the medicine?**



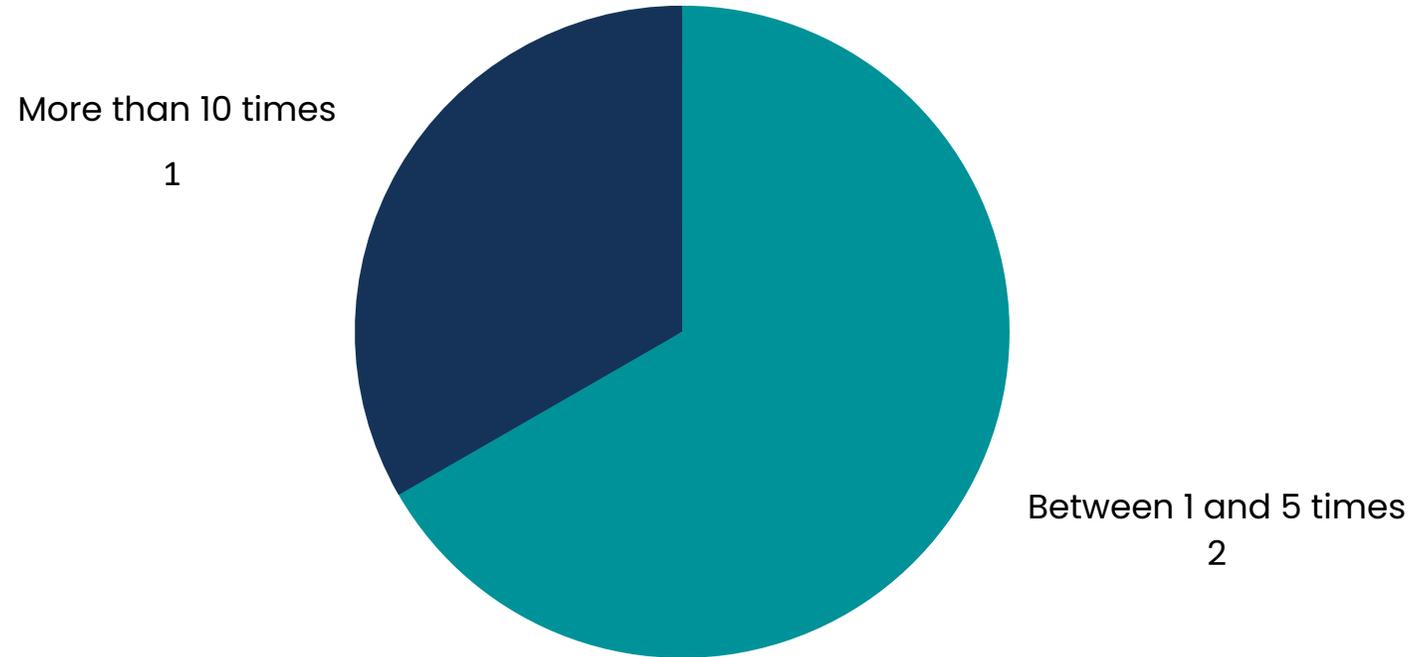
**13. Has the child/adolescent experienced any adverse effects during or after taking the medicine?**



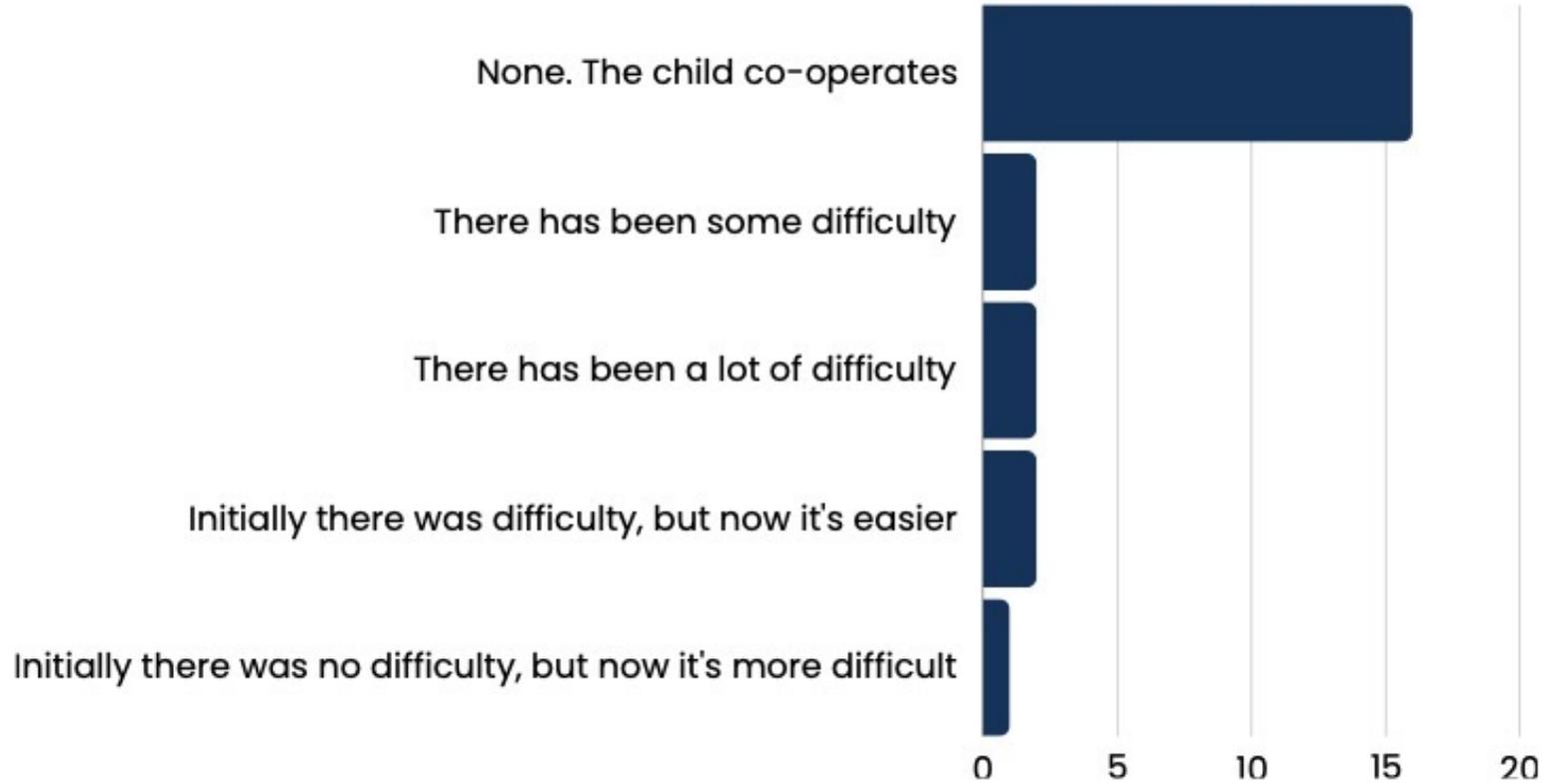
**14. What adverse effects has the child/adolescent experienced during or after taking the medicine?**



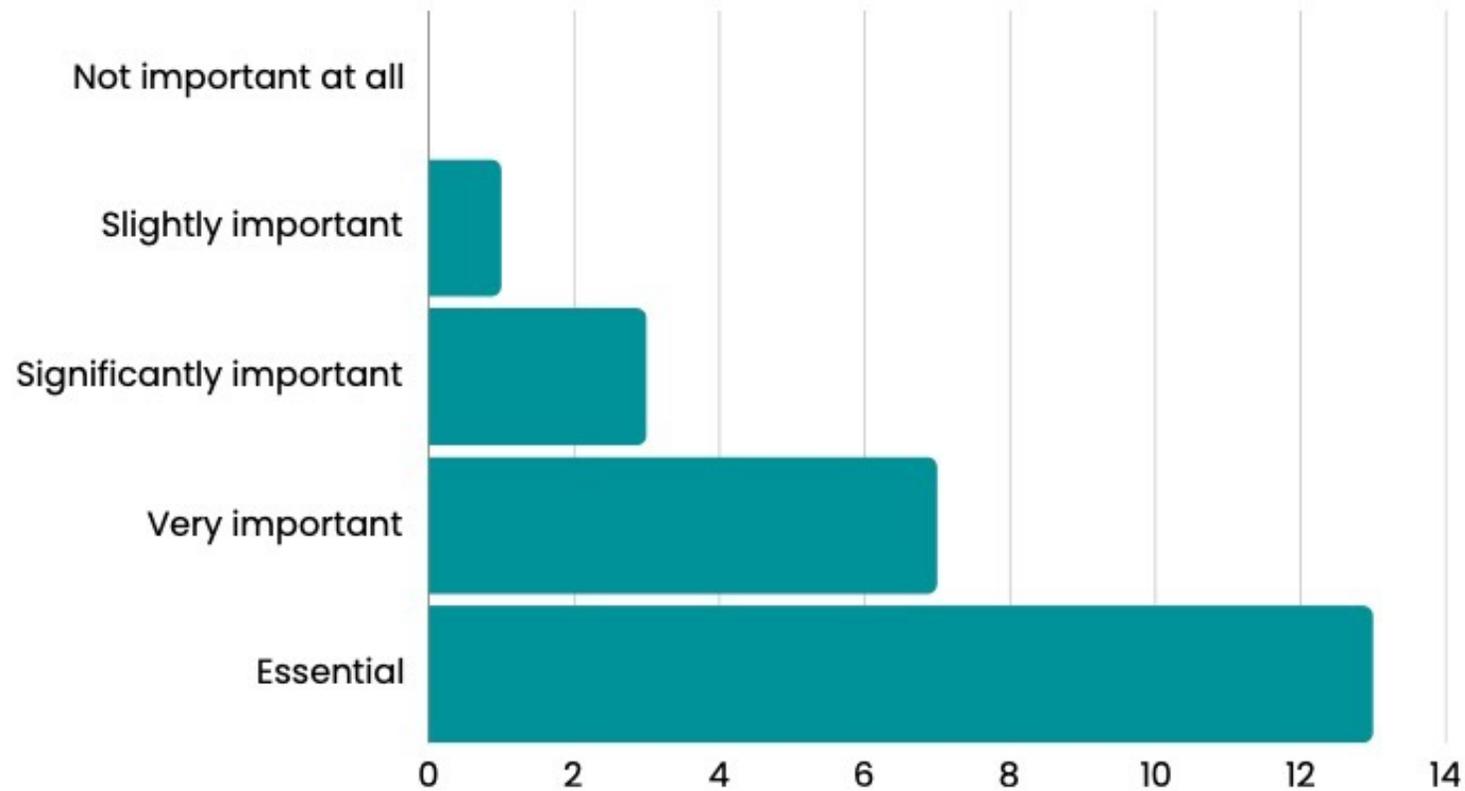
**15. How many times have adverse effects occurred since starting treatment?**



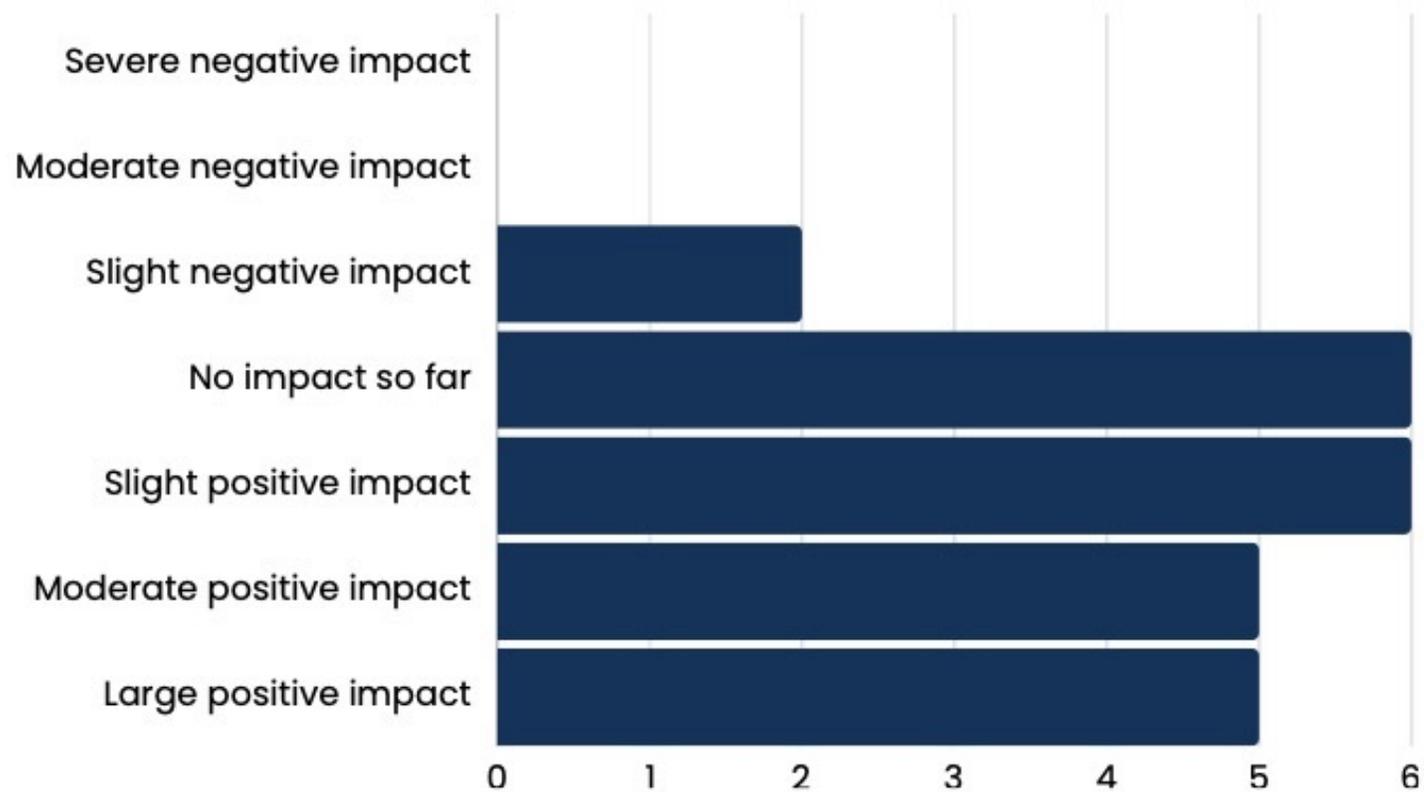
## 16. What challenges have you had with anthropometric measurements?



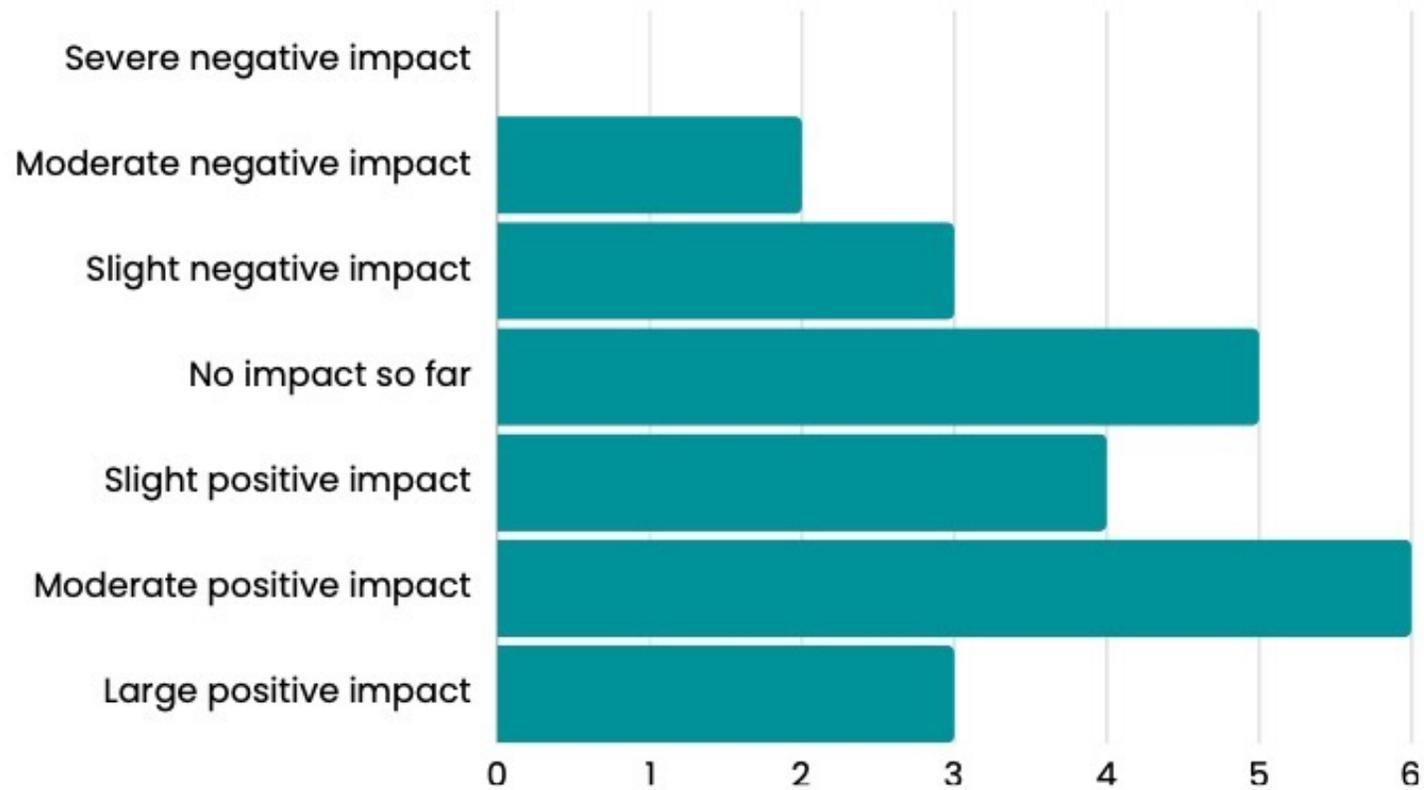
**17. How important do you think the consultations and clinical assessments associated with the treatment plan are for measuring treatment success?**



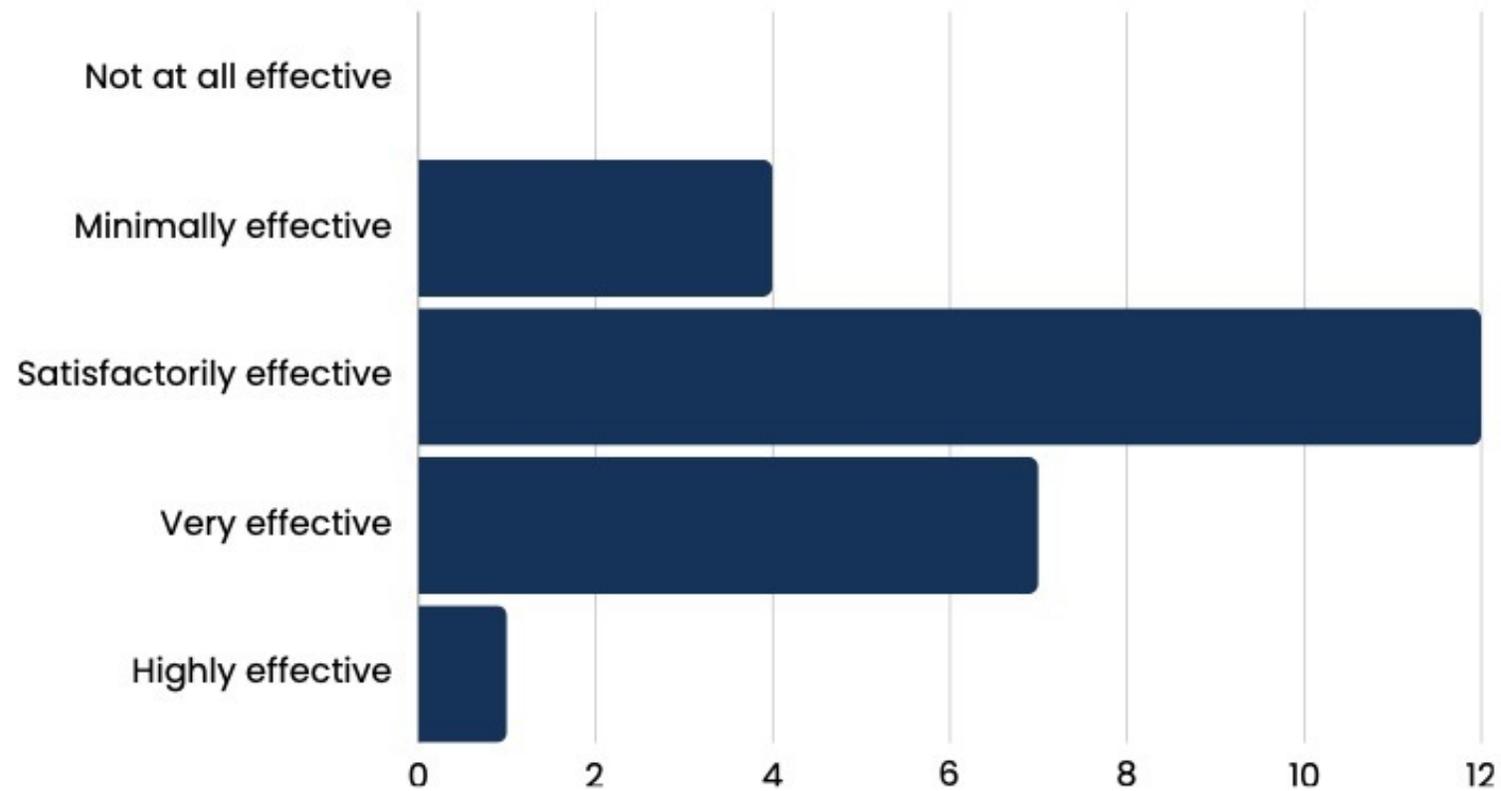
**18. In terms of the impact of treatment on the child's/adolescent's quality of life, do you consider that it had:**



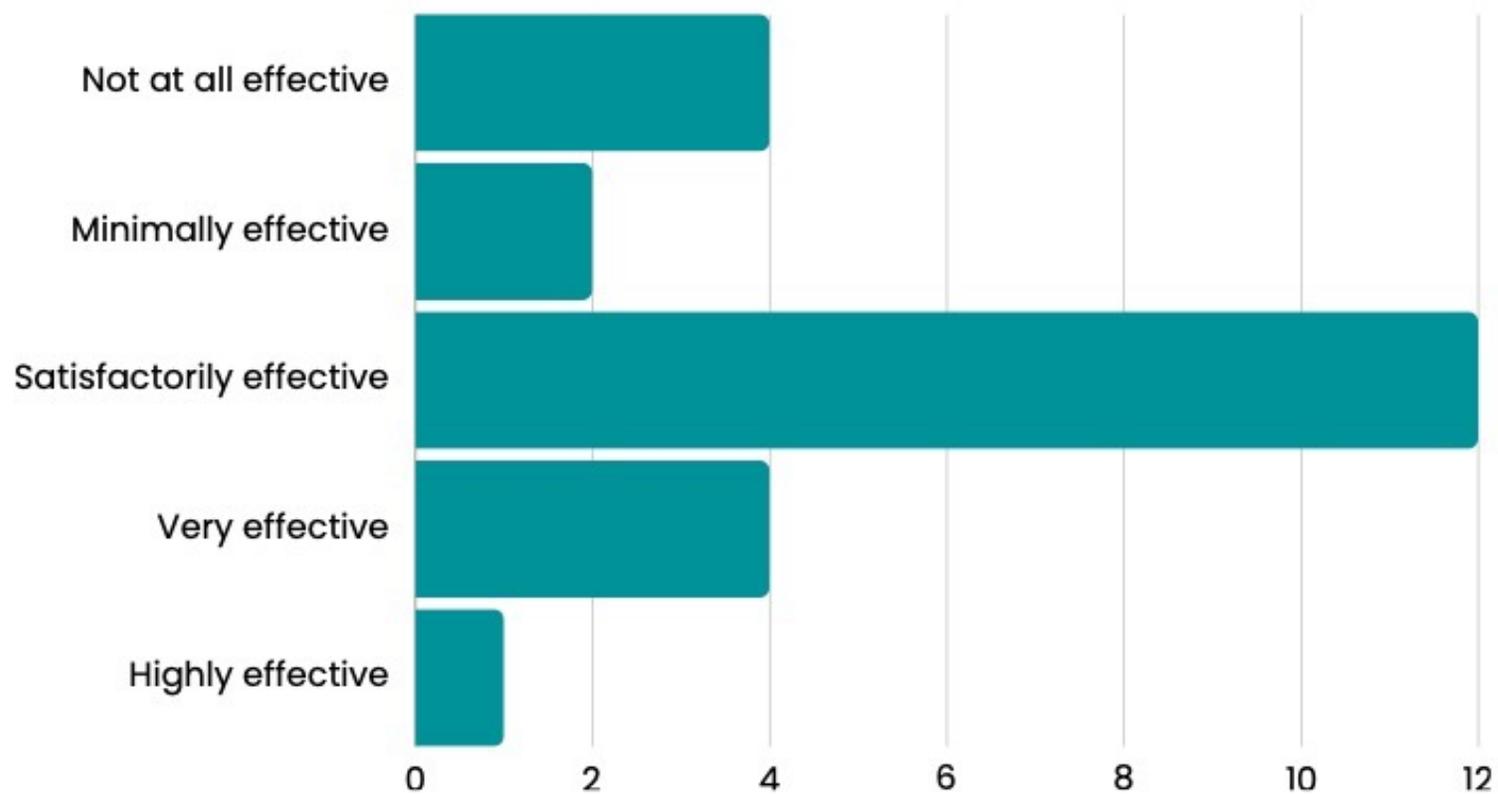
**19. In terms of the impact of treatment on the family's quality of life, you consider that it had:**



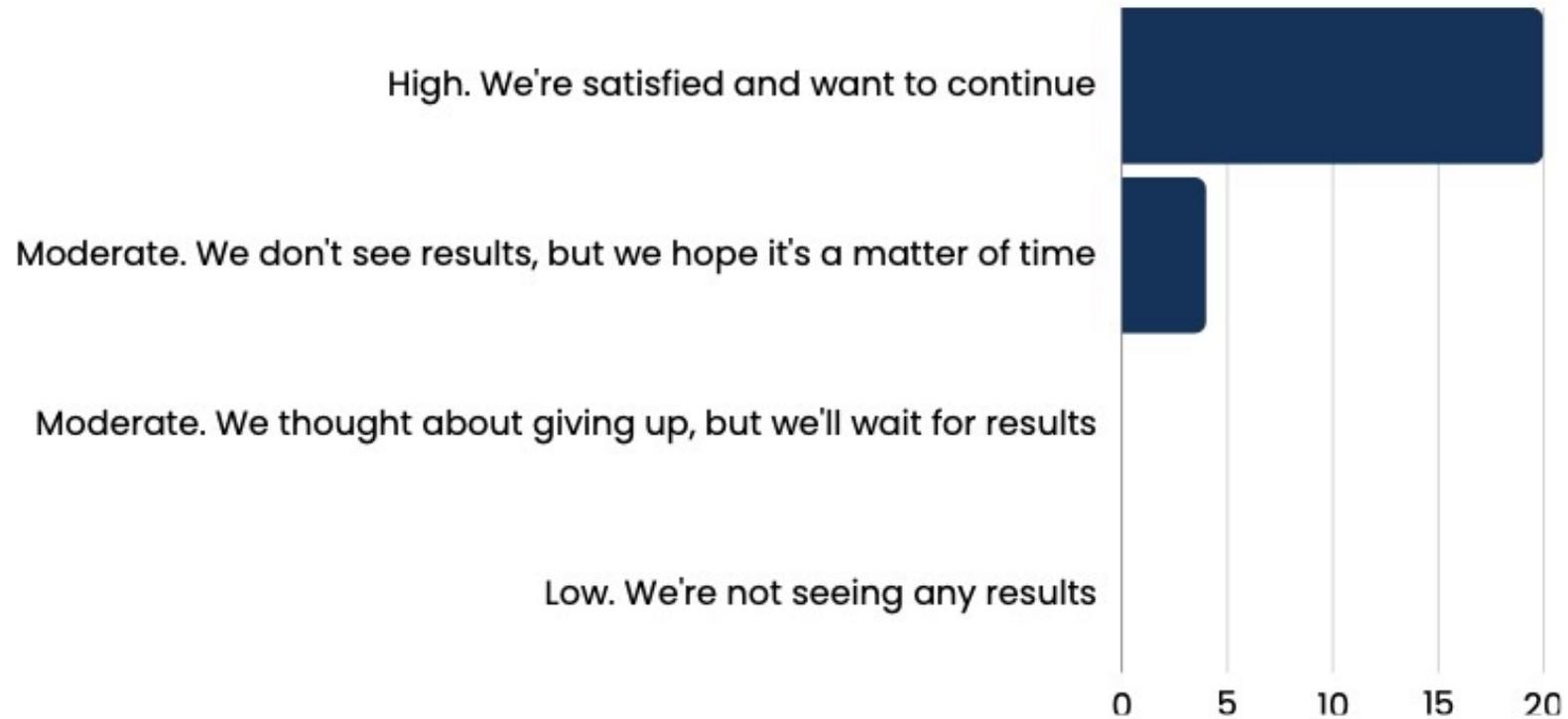
**20. Regarding your perception of the treatment's results on the child's/adolescent's bodily or physical development, to what extent do you think the treatment with this medicine is being effective?**



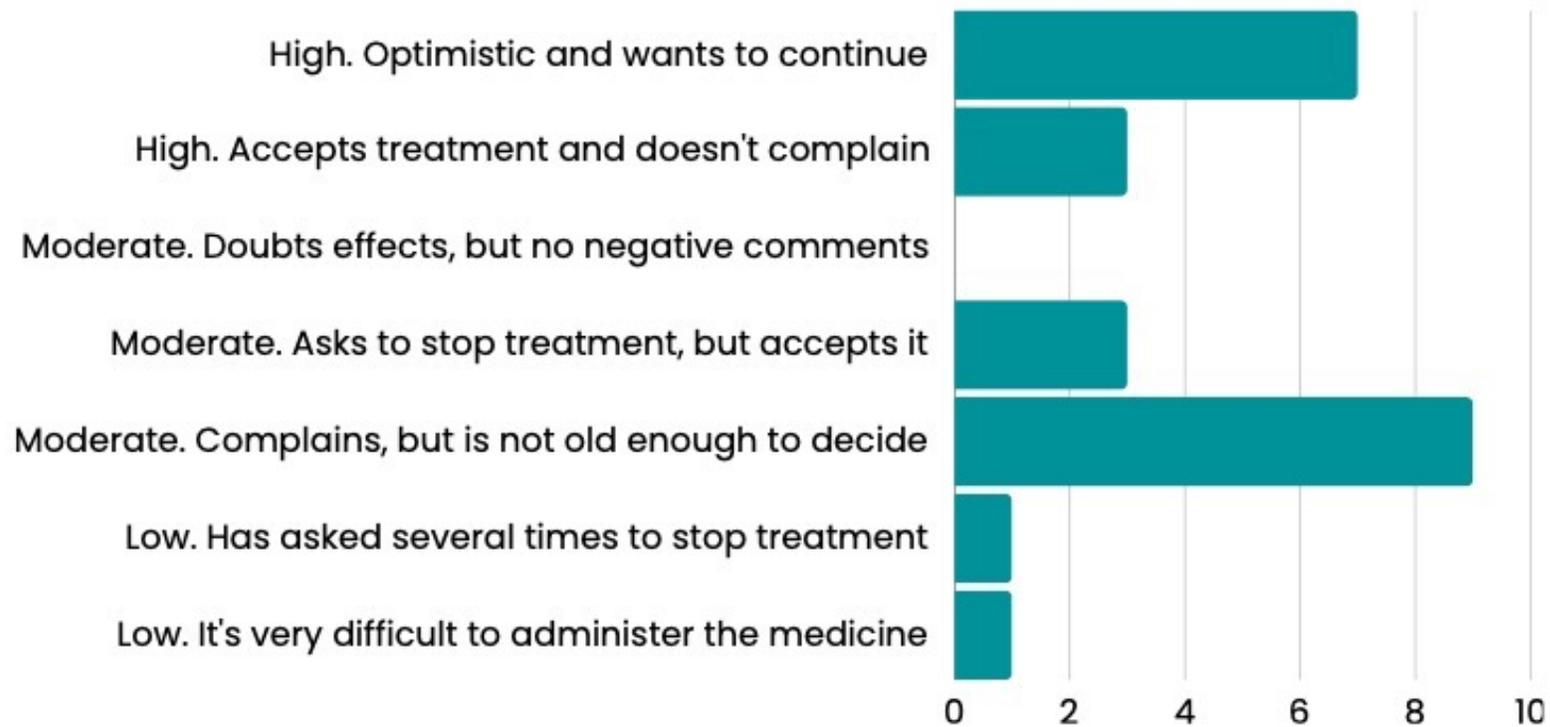
**21. Regarding your perception of the treatment's results on the child's/adolescent's psychological and emotional well-being, how effective do you think the treatment with this drug is being?**



**22. How motivated are parents to continue with their child/adolescent's current treatment?**



### 23. How motivated is the child/adolescent to continue with the current treatment?



# Doubts during the decision of starting therapy

- *Is it **worthy** do do a daily injection not being sure of the efficacy at the long-term?*
- *Is **is worthy** all this? Daily injection, monitoring, for 1-2 cm more per year...*
- *Not that many years left before growth will stop, **is it still worthy?***
- ***Short-term negative impact on quality of life vs long-term positive impact?***
- *What if in my child there will be **no effect** on growth?*
- ***Recent treatment**, limited information, and all the consequences of it...*
- *What if there will be **long-term side effects?***
- *Can the side effects turn out to be severe?*
- *Injection-related **pain***
- *Will my child continue to **refuse/resist the injection?***
- ***Will my child in the future agree** that we chose the treatment?*

# Main reasons for starting therapy

- **Better quality of life** for my child in the future!
- Improve **autonomy** in the future
- Improve **wellbeing** in the future
- Even if the improvement in growth is small, it will already be helpful
- Likely no need in the future for surgeries / limb-lengthening
- It is what my child wants!
- The clinical trials results are encouraging, the side effects were very mild
- It is the only treatment available
- Hope that will help in prevention of complications

Other question on present expectations, similar answers and: - **improve disproportion**

# Medical treatments for achondroplasia – just na option

- In our experience, most of the families in Portugal chose to do the treatment
- Reasons for families to choose not to do the treatment?
  - Examples:
    - Age group 12-15 years – joint decision: few years left of treatment – efficacy not as expected; teenager refusing injections; already wanting to do limb-lengthening;
    - Age group < 12 years: 2 cases (out of ~30)
      - *My child is doing well*
      - *We are afraid of the possible side effects*
      - *We do not want that our child to do any treatment for his dysplasia*

# Training for preparation and injections

Do you consider having had appropriate training to be able to do the injections at home?

- Yes, 100%
- Comments:
  - 2/24 mentioned that more training at the hospital would have been beneficial
  - One suggestion of doing a leaflet with tips/practical information
    - *The two-days-training at the hospital was crucial*
    - *Important on each visit to be able to discuss with nurses the evolution*
    - *Important to know the experience of other families*

# Fears

What are your main fears related to the treatment?

- **Non efficacy** / *no significant improvement in growth in my child*
- **Other non described side effects**
- *My child keeping refusing the injections and get **traumatized***
- *Daily injections leaving psychological sequela*
- *When my child becomes a teenager / adult, regrets having done the treatment, does not agree with our decision*
- *That my child in the future blames us of something*
- *Increases disproportion*
- *“Lack” of medication in the future*

Family video – for reviewers, please note this is a placeholder slide, the video will be reviewed as a separate job in Veeva

Questions?



# Quiz

Sérgio Sousa

Please hand in your quiz questions when you have completed the questions.

# Final discussion and closing remarks

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# Thank you!

Please join us for a coffee before your transfer back to the hotel.