The patient perspective on treatment for Skeletal Dysplasias

Sérgio B. Sousa and Alice Mirante
Ellie Simmonds: A World Without Dwarfism? - documentary explores views on new drug

By Claran 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.

https://www.youtube.com/watch?v=JblSfoyPJW4
https://www.bbc.com/sport/swimming/60934176
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
Questionnaire CHUC-ANDO Aug/SepP 2023

- Anonymous
- 32 replies
- 27 children with achondroplasia
- 24 on treatment
1. Age group of the child/adolescent

- < 2 years
- 2 - 4 years
- 5 - 8 years
- 9 - 11 years
- 12 - 14 years
- > 14 years
2. Sex of the child/adolescent

- Female: 10
- Male: 14
3. For how many months has the child/adolescent started their current treatment?

- < 6 months
- 6 - 12 months
- 12 - 24 months
- > 24 months
4. At what time of day/night does the child/adolescent take the medicine?

- Morning
- Afternoon
- Early evening
- Middle of the night
5. What factors influence the timing of taking the medicine?

- When parents are available
- Parents think it’s the best time
- We believe the time chosen increases efficacy
- The timing depends on when we can/the child lets us
- It’s mentioned in the leaflet
- By suggestion of health professionals
6. Who administers the medicine?

- Father
- Mother
- Mother and father alternate
- Other relative
- Child/adolescent themselves
7. Do you have to carry out any steps other than preparing the medicine?

- Apply analgesic to the injection site
- Prepare the room where the medicine is administered
- Prepare the child/adolescent
- I don't have to carry out any steps
- Other: apply ice before injection

0 2 4 6 8 10 12
8. Since starting treatment, state how many times you have missed a dose

- More than 10 times a month
- Between 5–10 times a month
- Between 2–5 times a month
- 1 time per month
- Between 1 and 4 failures in total
- Never missed a dose
9. Why did you miss the dose(s)?

- I forgot
- Refusal by the child/adolescent
- Run out of medication – not available at hospital
- Loss of medicine (bottle broken or tablet lost)
- Failure to deliver the medicine (transport)
- Lack of material to prepare the medicine
- Lack of painkiller
- Technical difficulties with administration
- Person needed for administration not present
- Illness of the child/adolescent
- Holidays / days off
- Past the administration time
- Difficulty in family planning
- Financial issues
10. Describe what you notice about the child/adolescent before the medicine is administered?

- Quiet
- Anxious
- Restless
- Hides
- Says "No"
- Asks why they have to take the medicine
- Complains or swears
- Other: cries
- Other: depends on how tired they are

0 2 4 6 8 10 12
11. Describe what you notice about the child/adolescent during the administration of the medicine?

- Quiet
- Relaxed
- Distracted
- Tenses muscles
- Tries to run away
- Shows pain (verbally, facial expressions)
- Kicking
- Cries
- Complains or swears
- Other: discomfort
12. Describe what you notice about the child/adolescent after the administration of the medicine?

- Quiet: 12
- Relaxed: 2
- Indifferent: 4
- Goes to play: 12
- Complains that it hurts: 6
- Cries: 2
- Complains or swears: 4
- Other: complains of itching: 0
13. Has the child/adolescent experienced any adverse effects during or after taking the medicine?

- Yes
- Maybe, not sure
- No
14. What adverse effects has the child/adolescent experienced during or after taking the medicine?

- Head spinning
- Haematoma at the injection site
- Bleeding at the injection site
- Redness or swelling at the injection site

0 0.5 1 1.5 2
15. How many times have adverse effects occurred since starting treatment?

- Between 1 and 5 times: 2
- More than 10 times: 1
16. What challenges have you had with anthropometric measurements?

- None. The child co-operates
- There has been some difficulty
- There has been a lot of difficulty
- Initially there was difficulty, but now it's easier
- Initially there was no difficulty, but now it's more difficult
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:

- Severe negative impact
- Moderate negative impact
- Slight negative impact
- No impact so far
- Slight positive impact
- Moderate positive impact
- Large positive impact
19. In terms of the impact of treatment on the family’s quality of life, you consider that it had:

- Severe negative impact
- Moderate negative impact
- Slight negative impact
- No impact so far
- Slight positive impact
- Moderate positive impact
- Large positive impact
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?

Not at all effective

Minimally effective

Satisfactorily effective

Very effective

Highly 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?

- Not at all effective
- Minimally effective
- Satisfactorily effective
- Very effective
- Highly effective

0 2 4 6 8 10 12
22. How motivated are parents to continue with their child/adolescent’s current treatment?

- High. We’re satisfied and want to continue
- Moderate. We don’t see results, but we hope it’s a matter of time
- Moderate. We thought about giving up, but we’ll wait for results
- Low. We’re not seeing any results
23. How motivated is the child/adolescent to continue with the current treatment?

- High. Optimistic and wants to continue
- High. Accepts treatment and doesn’t complain
- Moderate. Doubts effects, but no negative comments
- Moderate. Asks to stop treatment, but accepts it
- Moderate. Complains, but is not old enough to decide
- Low. Has asked several times to stop treatment
- Low. It’s very difficult to administer the medicine
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**

Questionnaire CHUC-ANDO SEP 2023 – 24 children on treatment
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

Sérgio Sousa and Alice Mirante
Thank you!

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