

ASBMR

The American Society for Bone and Mineral Research

INTRODUCTION

MEETING

Achondroplasia (ACH) is a skeletal dysplasia caused by a mutation at the fibroblast growth factor receptor 3 gene (FGFR3), a negative regulator of growth. The mutation increases FGFR3 signaling, altering proliferation and differentiation of chondrocytes leading to a limbs to torso disproportionality and short stature¹. Other physical characteristics include macrocephaly, lumbar lordosis, small trident hands, and small feet.

Human balance results from a complex integration of several sensory motor control systems as the visual, vestibular, somatosensory and musculoskeletal, needed to generate appropriate motor responses. It integrates parameters as position, velocity, and acceleration of body sway⁶. Most studies in achondroplasia are related to pediatrics and to medical complications, yet there is limited information on postural control and balance, which are relevant parameters for daily living tasks and for movements in physical activity⁷.

Body posture is controlled in a combination of open-loop (feedforward) and closed-loop (feedback) mechanisms, and the quality of posture control depends on the formation of a valid body schema and on the optimization of control loops⁸. Also, anticipatory and dynamic postural adjustments are needed for balance control and postural stabilization⁵. Sway is an important measure to analyze the deviations of the center of pressure (CoP), while Approximate Entropy (ApEn), is relevant to quantify temporal series regularity⁵.

Understanding the dynamics of postural control in adults with achondroplasia and identifying body sway tolerance limits that allow balance to be kept, are relevant research questions for future studies.

METHOD

Four women with achondroplasia were recruited for the pilot study. Their physical activity habits were surveyed using an investigator developed questionnaire with 6 questions. Two women stated having regular physical activity, at least 30 minutes 3 times per week (Active women-AC), also presenting better results in diverse physical tests while two women stated having no physical activity habits, presenting worst results in the same tests (non-active women – NAC).

Posturography evaluations using an in-ground force plate (Bertec[®], Columbus Ohio) were conducted to quantifying the body sway of participants in standing positions, based on CoP displacements. Participants were asked to stand over the force plate, in a hard surface, and remain as still as possible for a period of 30 seconds. The following situations and positions were requested to be performed:

- 1. Bipedal open eyes (OE)
- 2. Bipedal closed eyes (CE)
- 3. Unipedal right foot, open eyes (OE_uR)
- 4. Unipedal left foot, open eyes (OE_uL)

TRADITIONAL AND NONLINEAR ANALYSIS

The traditional variables analysed were the Anteroposterior (AP) and Mediolateral (ML) sway range and the Total Excursion (TOTEX), a sum of all AP and ML oscillations during the assessment time of 30 seconds in each of the four situations.

The Approximate entropy (ApEn) of the AP and ML sway was the selected nonlinear variable to analyze the level of adaptability to motion. As traditional and nonlinear variables tend to be inversely proportional⁹, we expect that the results follow this tendency.

Postural control pilot study of traditional and nonlinear variables in women with achondroplasia

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Table 1. Participants data

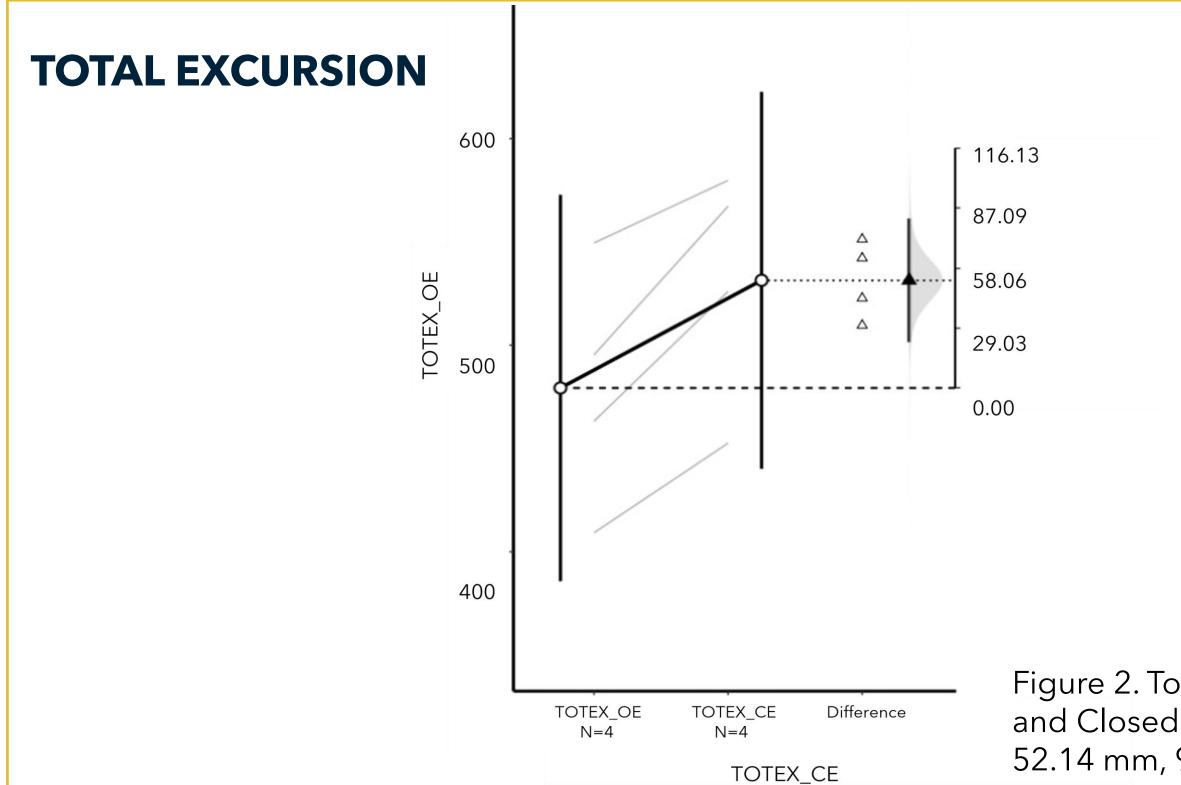
Active Women		Non Active Women
2	Participants	2
50±1.4	Age (years)	34.5±17.7
1.22±0.04	Height (m)	1.12±0.01
48.15±14.45	Weight (kg)	55.35±4.74
32.75±7.84	BMI (kg/m2)	44.05±4.17

AIM

Studies in the general population have demonstrated that an active lifestyle improves postural control² while balance deficits are frequently observed in people with musculoskeletal conditions³. This pilot study was conducted to assess postural control and balance in women with achondroplasia and possible correlations between physical activity habits and ability, body composition and biomechanical analysis. In this exploratory research, we have hypothesized that physically active women with achondroplasia have an improved postural control and increased adaptability to adjust posture and maintain balance.



over a force plate, hard surface



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Figure 1. Participant in bipedal stance, closed eyes (A) and in unipedal right foot, open eyes (B),

Figure 2. Total excursion for Open and Closed eyes (OE-CE). M. diff 52.14 mm, 95% CI [22.192-82.085]

RESULTS

We have conducted a descriptive analysis using Jamovi (Version 1.6 Sydney, Australia). Comparing the bipedal stance in Closed eyes and Open eyes, the first presented an increased sway and entropy as this is a more challenging situation. Comparing to Non-active women (NAC), the Active women (AC) presented an increased length for TOTEX in Closed eyes [553±38.0 vs 510±81.1] and Open eyes [506±61.1 vs 452±60.9], which is related to a higher ability to sway, suggesting increased postural control for steadiness. AC presented an inferior sway range at the Anteroposterior (AP) component in Closed eyes [17.3±1.09 vs18.3±2.25] and Open eyes [9.70±1.38 vs 15.0±4.25] as well for Mediolateral (ML) component in Closed eyes [29.8±1.28 vs 31.4±10.2] as in Open eyes [22.6±4.9 vs 22.8±11.2], having inferior displacements yet this does not inform about the quality of the sway. Analyzing the Approximate Entropy, AC presented higher values at the AP component, in Closed and Open eyes [0.826±0.101 vs 0.630±0.122; 0.888±0.1 vs 0.699±0.2] and as well for the ML component, also in Closed and Open eyes [0.550±0.01 vs 0.535 ± 0.1 ; 0.583 ± 0.1 vs 0.569 ± 0.2] suggesting a better capacity to adapt to irregular and unpredictable external disturbances. Although it is expected that at an unipedal stance the sensory motor systems are more challenged, the data collected don't allow us to see posture tendencies. Yet, while one NAC participant had a dominant left foot, both AC had the right dominant foot and presented increased sway range at unipedal left.

POSTURAL CONTROL ANALYSIS Table 2. Mean values and SD (mm) of total excursion, sway and approximate entropy. AC- Active Women; NAC - Non-active women											
	Total excursion (mm)		Anteroposterior Sway range (mm)		Mediolateral Sway range (mm)		Anteroposterior Approximate entropy		Mediolateral Approximate entropy		
	AC	NAC	AC	NAC	AC	NAC	AC	NAC	AC	NAC	
Bipedal Closed eyed	553±38.0	510±81.1	17.3±1.09	18.3±2.25	29.8±1.28	31.4±10.2	0.826±0.10	0.630±0.12	0.550±0.01	0.535±0.11	
Bipedal Open eyes (OE)	506±61.1	452±60.9	9.70±1.38	15.0±4.25	22.6±4.9	22.8±11.2	0.888±0.12	0.699±0.22	0.583±0.13	0.569±0.22	
Unipedal right OE	902±210	1190±19.8			29.5±2.07	50.2±10.0			0.625±0.08	0.661±0.05	
Unipedal left OE	1059±432	1130±655			64.1±28.60	47.2±8.16			0.543±0.09	0.605±0.13	

CONCLUSIONS

Based on these preliminary results, there is a tendency for physically active women with achondroplasia to have increased postural control and adaptability to modulate postural changes in comparison with non-active women, who tend to show more robotic movements to keep postural control. Study limitations include the reduced number of participants and lack of balance losses record. Although no preliminary conclusions can be taken from the unipedal stance, further assessments may be useful to better understand balance and sensory motor skills. Future studies will be conducted to analyze movement in adults with achondroplasia, identify postural control strategies and associations between physical and biomechanical parameters.

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