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Facilitators and constraints of physical activity in adults with achondroplasia: a scoping review

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Introduction

Achondroplasia (OMIM no. 100800), a skeletal dysplasia, is caused by a mutation in the fibroblast growth factor receptor 3 gene (FGFR3), leading to abnormalities in bone growth and modelling [10, 49]. With a global prevalence of 1 in 25,000 births [19], achondroplasia is characterized by disproportional short limbs and an adult height around 1.3 m [36]. Individuals with achondroplasia may experience various medical complications, such as obesity [53], spinal stenosis [21], joint hyperlaxity, early-onset arthritis, restricted pulmonary function [20], and reduced exercise capacity [14]. The phenotype variability is substantial and attributed to environmental and genetic factors [45]. Most adults with achondroplasia are autonomous, although some require adjustments to activities of daily living [32, 54]. The distinctive appearance also poses psychological challenges and social stigma [61], with minimal data regarding the social experience of living with this condition [61]. Physical activity includes all bodily movements produced by skeletal muscles that result in energy expenditure [7]. Exercise, a subset of physical activity, is planned, structured, and repeated to improve or maintain physical fitness [7], which is the body ability to adapt to the environment and perform daily activities without excessive fatigue [47]. Despite well-documented benefits of physical activity and fitness for the general population, data in achondroplasia is limited, and the facilitators and constraints of physical activity in this population remain unclear. Some interventions, such as tailored strength and balance training programs, may lead to improvements in physiological and biomechanical outcomes [14, 57, 58] and participation in adapted sports participation [17, 29, 50], although randomized trials are lacking [34]. This scoping review aimed to (a) identify physical, physiologic, biomechanical, psychologic, and nutritional factors that may facilitate or constrain physical activity, exercise, and adapted sports in adult individuals with achondroplasia and (b) explore how these factors may influence physical activity motivation, participation, and capacity in this population.

Methods

We used the Cornell University Library decision tree [38] to determine the most appropriate literature review type for our broad research question on physical activity among adults with achondroplasia. We selected the

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scoping review methodology and followed the guidance from the Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) [62]. We searched major databases, supplemented with manual searches of key journals and gray literature sources. This scoping review protocol is registered with the Open Science Framework repository, https://osf.io/gnhpc. We used the NIH Quality Assessment Tool [46] and the Appraisal of Guidelines for REsearch & Evaluation tool (AGREE II) [4] for the quality appraisal of the included records.

Eligibility criteria

To capture multidimensional factors related to physical activity, we included all studies found approaching motion analysis, biomechanics, gait, strength, functional movements, physical activity and fitness, exercise, adapted sports, body composition, and psychosocial and nutritional aspects in people with achondroplasia over 18 years. All study designs were eligible, including quantitative, qualitative, and mixed methods, as well as abstracts and gray literature, from any publication year and language. Studies including participants with achondroplasia and with other conditions were also included. We excluded studies focusing on genetics, surgery, pharmacologic treatments, pediatric populations, animal models, or solely other skeletal dysplasias, as well as records with unavailable full texts.

Information sources and search strategy

To obtain relevant records, we searched six databases (PubMed, Embase, Ovid, MEDLINE, CINAHL, and PsycINFO) in October 2022 and identified additional records through manual searching. Pre-defined search terms included the following: achondroplasia, movement, physical activity, exercise, sport, body composition, biomechanics, kinetics, kinematics, electromyography, mental health, and nutrition. All possible combinations of these search terms were entered as separate queries and combined the results. An experienced librarian drafted the search strategies, which were refined through team discussion. The final MEDLINE search strategy is presented in Table 1.

Selection of sources of evidence and data charting

The search records were downloaded into reference management software EndNote 20. The eligibility criteria were piloted through an initial discussion with topic experts, to reduce ambiguity. Two researchers (I. A. and F. K.) independently screened all titles and abstracts against the predefined eligibility criteria, including records with relevant titles even without and abstract. Disagreements were resolved through joint full-text review and discussion between, with a third researcher (F. R.) arbitrating if needed. Records with unclear participant age underwent full-text review. We identified additional eligible records through manual searching. Both researchers reviewed all retrieved full texts to confirm final inclusion status using the same consensus process.

Results

Selection and characteristics of sources of evidence

Our search returned 811 records. After excluding duplicates ($n = 291$), two reviewers screened 520 records by title and abstract, resulting in a selection of 52 records. Among these, 8 were excluded (3 for being pediatric studies and 5 for the inability to retrieve full texts) resulting with a selection of only 44 eligible records. A manual search was also conducted in the following weeks, retrieving 6 records more, which resulted in a final number of 50 records included in this review. The screening process is documented in a PRISMA-ScR study flow diagram (Fig 1).

Table 1 Sample Embase search strategy summary

Combined search terms usedCombined search terms used	'achondroplasia'/de OR (achondroplas*):ab,ti,kw) **AND** ('physical activity'/exp OR 'exercise'/exp OR 'physiotherapy'/exp OR 'kinesiotherapy'/exp OR 'body composition'/exp OR 'lean mass'/de OR 'kinetics'/exp OR 'electromyography'/de OR 'surface electromyography'/de OR 'sport'/exp OR 'mental health'/exp OR 'mental disease'/de OR 'wellbeing'/exp OR 'daily life activity'/de OR 'social life'/de OR 'social behavior'/de OR 'social stigma'/de OR 'nutritional health'/de OR 'diet'/exp OR 'dietary intake'/exp OR 'diet therapy'/exp OR 'dietary pattern'/exp OR 'food frequency questionnaire'/de OR 'nutrition'/de OR 'food intake'/de OR 'movement (physiology)'/exp OR (exercis* OR biomechanic* OR physiotherapy* OR kinesiotherap* OR ((physical* OR kines*) NEAR/3 therap*) OR ((physical*) NEAR/3 (activit* OR workout* OR work-out*)) OR body-fat* OR body-compos* OR lean-mass* OR motion* OR gait* OR locomotion* OR kinetic* OR kinematic* OR walk* OR electromyogra* OR sport* OR athlet* OR ((mental* OR emotional*) NEAR/3 (health* OR disorder*)) OR psycholog* OR psychopatholog* OR well-being* OR wellbeing* OR ((daily*) NEAR/3 (activit* OR function*)) OR social* OR stigma* OR nutrition* OR diet* OR (motor* NEAR/3 (assess* OR behaviour* OR behavior*)) OR movement* OR mobilit*):ab,ti,kw) *NOT ('juvenile'/exp NOT 'adult'/exp)*

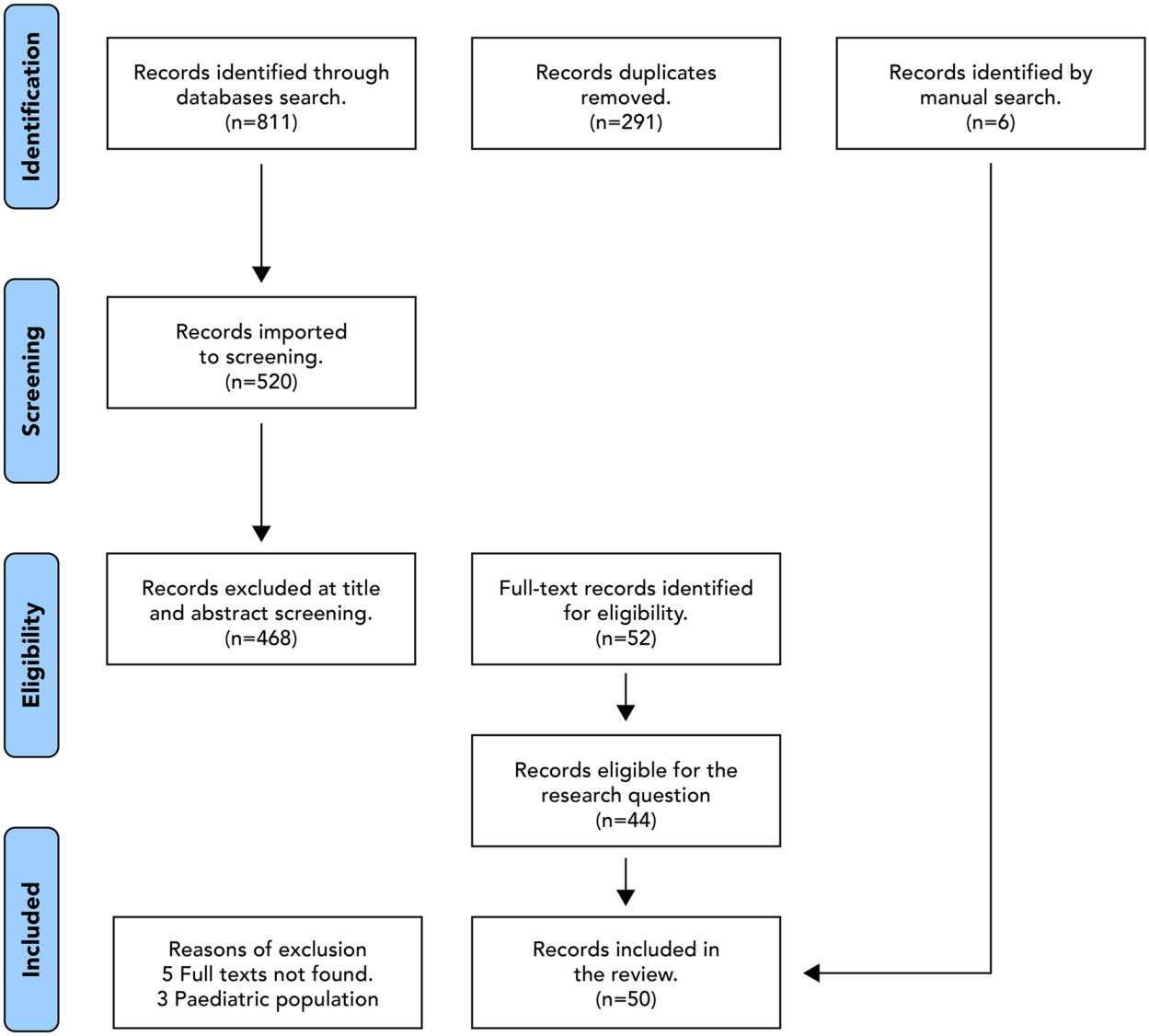

Fig. 1 PRISMA-ScR flow diagram

Relevant characteristics of the selected sources, as nature and extent of involvement of advocacy organizations in the selected sources on study design, conduct and analysis, and recruitment, are presented in Table 2. The evidence base consisted primarily of 30 cross-sectional studies (60%), followed by 5 literature reviews (10%), 5 case reports (10%), 2 mixed-methods studies (4%), 2 expert commentaries (4%), 2 perspective papers (4%), 1 illustrative qualitative study (2%), 1 retrospective observational (2%), 1 symposium proceedings (2%), and 1 guideline (2%). The included sources had a range of sample sizes, from single case studies to large cross-sectional surveys with over 400 participants. In total, the sources included data on 1997 participants with a diagnosis of achondroplasia, with an age range between 8 and 84 years. The most common topics of the sources was on quality of life and health status ($n = 12$), followed by body composition ($n = 7$), biomechanics/gait analysis ($n = 7$), and physical activity levels, capacity, or interventions ($n = 5$). The most frequently used outcome measures were questionnaires ($n = 18$), anthropometrics ($n = 6$), cardiopulmonary exercise testing ($n = 3$), and gait analysis ($n = 3$). Among the 50 included sources, only 7 involved participation of advocacy groups at different levels, including study design, conduct, analysis, and co-authorship of sources.

The quality of the included sources was assessed using the NIH Quality Assessment Tools and the AGREE II

Table 2 Main characteristics of the included sources and advocacy organizations involvement in research

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
Bailey, J. et al.	1971	Retrospective observational	41	41	0 to 72	Upper limb deformities	Prevalence of radiographic elbow abnormalities	x	x
Brust, S. et al.	1976	Cross-sectional	16	11	19 to 80	Psychiatric aspects	Psychiatric interviews, psychological tests	x	x
Hooks, M. et al.	1986	Case report	1	1	22	Nutritional support and estimating caloric need	Anthropometrics, biochemical tests, indirect calorimetry	x	x
Hecht, J. et al.	1988	Cross-sectional	196 +241 (records)	196 + 241 (records)	0 to 70	Obesity	Anthropometry, body mass index, skinfold thickness	x	LPA
Hall, J. et al.	1989	Conference proceedings	x	x	x	Multidisciplinary	NA	x	LPA participation
Owen, O. et al.	1990	Cross-sectional	32	27	18 to 54	Resting metabolic rate and body composition	Resting metabolic rate, body composition	x	LPA and LP Canada
Low, L. et al.	1996	Literature review	x	x	x	Structural, developmental, medical, and orthopedic characteristics	NA	x	x
Apajasalo, M. et al.	1998	Cross-sectional survey	121	8	16 to 54	HRQoL	15 D and 16 D multidimensional quality-of-life questionnaires	x	x
Mohamed, N. et al.	1998	Cross-sectional survey	1500	437	Mean 38	Functional health status of adults	SF-36 Health Survey	x	LPA distributed the survey
Gollust, S. et al.	2003	Cross-sectional survey	325	189	mean 41	QoL	Ferrans and Powers Quality-of-Life Index (QLI)	x	LPA Medical Advisory Board
Carneiro, J. et al.	2007	Case report	1	1	28	Obesity and bariatric surgery	6-min walk test, SF-36	x	x
Cervan, M. et al.	2008	Cross-sectional case control	42	21	14 to 53	QoL	WHOQOL-BREF questionnaire	x	AGPB
Thompson, S. et al.	2008	Literature review	x	x	x	Psychosocial and medical factors	NA	x	x

Table 2 (continued)

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
Cortinovis, I. et al.	2011	Mixed-methods research	18	18	23 to 48	Daily life and experience	Experience sampling method, flow questionnaire, Life Theme Questionnaire	One AISAC member is co-author	AISAC
Dlugash, R. et al.	2011	Cross-sectional	3	3	34 to 44	Energy balance	Dietary intake, resting energy expenditure, physical activity expenditure	x	x
Castro, E. et al.	2012	Case report	2	1	28	Gait analysis	Gait velocity, cadence, step length, stride length, stride duration	x	x
Henry, B. et al.	2012a	Methodological Ccross-sectional	20	20	Not specified	Anthropometric measurements	Dual-energy X-ray absorptiometry (DXA), bioelectrical impedance analysis (BIA), and anthropometry	x	x
Henry, B. et al.	2012b	Cross-sectional	19	19	Not specified	Diet and nutrients intake	Diet records + comparison with (WWEIA 2007-2008)	x	x
Schulze, K. et al.	2012	Cross-sectional	20	20	Not specified	Body composition	Anthropometry, dual-energy X-ray absorptiometry (DEXA)	x	LPA
Schulze, K. et al.	2013	Commentary/ perspective. theoretical analysis	x	x	x	BMI in achondroplasia	NA	x	x
Chiriti, G. et al.	2014	Case report		1	42	Physical and kinetics rehabilitation	VAS pain score, Tinetti Gait Scale score, ADL score	x	x
Hoobs, D.	2014	Review article	x	x	x	Training athletes with achondroplasia	NA	x	x

Table 2 (continued)

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
DSAUK	2015	Qualitative study using illustrative case studies	8	Unspecified	17 to 28	Sports participation	Semi-structured interviews	Unclear	DSAUK
Rodriguez-Gomez, J. et al.	2015	Cross-sectional	22	22	21 to 75	Psychosocial and medical factors	Beck Depression Inventory, Beck Anxiety Inventory, Beck Hopelessness Scale, Symptom Checklist-90-Revised	x	LPPR
Rohenkohl, A. et al.	2015	Cross-sectional survey	89	33	18 to 28	QoL and psychological health	KIDSCREEN-10, DISABKIDS, QoLISSY questionnaire, and Strengths and Difficulties Questionnaire (SDQ)	BKMF cooperated in study conduct	BKMF
Dhiman, N. et al.	2017	Cross-sectional survey	189	106	19 to 80	HRQoL	SF-12 questionnaire on health-related quality of life	x	LPA
Etayo, L.	2017	Commentary paper	x	x	x	Nutritional challenges	NA	x	x
Gomez, E. et al.	2017	Cross-sectional with participatory research	45	45	21 to 46	Footwear design	Survey by researchers + antropometric and baropodometric equipment	x	x
Da Rocha, L. et al.	2018	Mixed-methods research	8	8	Over 18	Psychosocial aspects and inclusion and physical activity	Semi-structured interview, FMS test	x	x
Matsushita, M. et al.	2018	Cross-sectional	201	Unspecified	10 to 67	HRQoL	SF-36 Health Survey	x	GTA
Sims, D. et al.	2018	Cross-sectional	27	10	Mean 22	O2 consumption and gait metabolic cost	Portable indirect calorimetry	x	DSAUK

Table 2 (continued)

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
Jennings, S. et al.	2019	Cross-sectional survey	336	192	18 to 81	Social inclusion	PHQ-8, GAD-7, Brief Pain Inventory	x	LPA
Madsen, A. et al.	2019	Cross-sectional, descriptive	33	33	Mean 40	Anthropometrics, diet, and resting energy expenditure	Anthropometrics, SmartDiet questionnaire, dietary records, indirect calorimetry, and predictive equations.	6 members of NIK participated in study protocol, selecting main topics, piloting questionnaires	NIK
Matsushita, M. et al.	2019	Cross-sectional survey	201	108	18 to 67	HRQoL	SF-36 questionnaire	x	GTA
Sims, D. et al.	2019a	Cross-sectional	27	10	mean 22	Body composition analysis	Anthropometry, dual-energy X-ray absorptiometry (DEXA)	x	DSAUK
Sims, D. et al.	2019b	Cross-sectional	27	10	Mean 22	Gait analysis	3D motion analysis and electromyography	x	DSAUK
Gattlieb, S. et al.	2020	Quantitative experiment	101	101	25 to 65	Social dominance orientation and discrimination	Job Suitability Scale, Decision to Interview Scale, Social Dominance Orientation Scale	x	x
Sims, D. et al.	2020	Cross-sectional	27	10	Mean 22	Gait analysis	3D lower limb joint kinematics. Standard Plug-in-Gait model	x	DSAUK
Constantinides, C. et al.	2021	Literature review	x	x	x	QoL, physical functioning, and psychosocial function	NA	x	x
de Vries, O. et al.	2021	Cross-sectional	43	43	Mean 40	Physical fitness	VO2peak, 6MWT, 30sSTS, BESS, IPAQ	6 members of NIK participated in study protocol, selecting main topics, piloting questionnaires	NIK

Table 2 (continued)

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
Fredwall, S. et al.	2021	Cross-sectional case control	49	49	Mean 41	Cardiovascular risk factors and body composition	Anthropometrics, blood pressure, cholesterol, glucose, HbA1c, MRI for body composition	6 members of NIK participated in study protocol, selecting main topics, piloting questionnaires	NIK
Hoover-Fong, J. et al.	2021	Expert review and perspectives	x	x	x	Natural history of achondroplasia	NA	x	x
Ireland, P. et al.	2021	Cross-sectional	150	25	21 to 75	Screening Tool for Everyday Mobility and Symptoms validation	STEMS, Functional Mobility Scale (FMS) and 6-minute walk test (6MWT)	x	SSPA
Maghnie, M. et al.	2021	Cross-sectional	186	74	18 to 84	HRQoL	EuroQoL 5 dimensions questionnaire, Brief Pain Inventory (short form) (BPI-SF), Nottingham Health Profile part I (NHP)	x	x
Munoz-Reyes, F. et al.	2021	Cross-sectional	48	Unspecified	Unknown	Body proportions and composition	Anthropometric dimensions. No details provided on specific standardized tools	x	x
Rincón Rueda, Z. et al.	2021	Case report	1	1	27	Physiotherapy treatment plan	Aerobic capacity (modified Bruce protocol), joint ROM, muscle strength, posture, gait, pain (VAS)	x	x
Yonko, E. et al.	2021	Retrospective review	25	25	19 to 66	QoL	SF-36 Health Survey. Psychiatric illness diagnoses	x	x
Ajimi, A. et al.	2022	Cross-sectional	86	78	> 20	Daily living	Open-ended questionnnaire on inconveniences and adaptations	x	x

Table 2 (continued)

Author	Publication year	Study design	Sample size	Number of adults with ACH	Age range	Primary topic	Outcome measures	Advocacy groups involvement in study design, conduct, and analysis	Advocacy groups support in recruitment
Jacinto, M. et al.	2022	Narrative review	x	x	x	Physical activity in skeletal dysplasias	NA	2 members of ANDO Portugal participated in the review design, conduct, and writing	x
Savarirayan, R. et al.	2022	Expert consensus guideline	x	x	x	Recommendations for management of achondroplasia	NA	3 patient organizations representatives were co-authors	x

Abbreviations: *LP* Little people, *LPA* Little People of America, *AGPB* Small People Association Brazil, *AISAC* Association for the Study of Achondroplasia, *DSAUK* Dwarf Sport Association United Kingdom, *BKMF* German Short Stature Association, *NIK* Norway short stature, *SSPA* Short Statured People's Association of Australia, *LLPR* Little People Puerto Rico, patient organization in Japan, *GTA* Glory to Achondroplasia, *ANDO Portugal* National Association for Skeletal Dysplasias

tool for one guideline. Risk of bias, study limitations, and quality rating for each publication were evaluated by two independent reviewers and discrepancies resolved through discussion. Overall, quality was categorized as good, fair, or poor based on the risk of bias, with good quality studies having the lowest risk of bias and poor-quality studies having the highest risk of bias. Studies where a quality assessment was not applicable, such as conference abstracts and qualitative interview reports, were denoted as not applicable (NA). The results of the quality appraisal for each included study are summarized in Table 3. Of the 50 sources included, 14 were rated as good quality, 24 as fair quality, 9 as poor quality, and 1 guideline quality was rated with a 5 (being7 the highest score), and in 2, quality assessment was not applicable (NA) as one was a conference proceeding and the other an illustrative cases study. Among the six reviews, three were high-quality systematic reviews, while the others provided narrative expert syntheses prone to potential selection biases in article inclusion.

Synthesis of results

Only 26% of the selected sources were published before 2009, as presented in Fig. 2. After 2010,research and development of innovative medicines achondroplasia started, with most publications focused on the pediatric population. Yet, there is still a void on high level of evidence research, specifically on randomized controlled studies.

The most relevant conclusions from the included sources provide insights into factors impacting physical activity participation among adults with achondroplasia, as presented in Table 4. Of the 50 included sources, 31 addressed issues directly related to physical activity capacity or motivation; 15 highlighted reduced physical functioning and physical quality of life compared to average stature individuals; 13 emphasized biomechanical limitations and gait alterations that may constrain exercise; 12 noted high rates of pain, chronic health conditions, and obesity that can limit activity tolerance; and 10 suggested adapted exercise programs, and sports participation may provide physical and psychosocial benefits.

The main findings were organized in four sections: physical and physiologic factors, biomechanical factors, psychological and psychosocial factors, and nutritional factors. Based on these findings, it was possible to identify facilitators and constrains of physical activity, exercise, and adapted sports for adults in achondroplasia, which are presented in the "Facilitators of physical activity, exercise, and adapted sports" section. From the 50 sources, 29 were relevant on physical and physiological factors, 4 on biomechanical factors, 12 on psychological and psychosocial factors, and 5 on nutritional factors. Regrading distribution of sources origin, 19 are from researchers based in North America, 15 from Europe, 7 from South and Central America, 5 are internationals, 3 from Asia, and 1 from Australia.

Discussion

The selected sources provide useful data on the epidemiology, diagnosis, management, and psychosocial aspects of achondroplasia. The cross-sectional studies provide useful insights into factors related to physical activity but most only through associative conclusions given their inability to control for confounders. Regarding the guideline included in this scoping review, a robust method (modified Delphi process) was applied, with limitations being the lack of systematic evidence review and reliance more on expert opinion. The few interventions studies were mostly pilot or preliminary investigations with methodological limitations. The case reports and series were limited by lack of controls and small samples. The preponderance of studies with fair or poor ratings highlights that more rigorous controlled research is needed across study designs to elucidate constraints on physical activity participation for adults with achondroplasia. Despite these limitations, several information was found on factors that constrain or facilitate physical activity, exercise, and sports for adults with achondroplasia, which are presented in the following sections.

Physical and physiological factors

Individuals with achondroplasia have disproportionately short stature and skeletal deformities due to impaired bone growth, resulting in rhizomelic shortening of the extremities, especially the proximal upper arm and thigh segments [31, 49]. These anatomical and physiological challenges can impair physical function, affecting overall quality of life [33, 42, 43]. Joint laxity and deformities are common, leading to limited ranges of motion and increased injury risk [29]. The hands show an inability to fully extend the fingers and have variable finger lengths, while the forearms have limited supination and lack of full elbow extension, which can reduce arm and hand function over time [3]. Body composition differs in achondroplasia, with lower total bone mineral content, bone density, and fat-free mass compared to average stature adults [55, 57, 58]. Adiposity and obesity are common in achondroplasia, but standard BMI categorization overestimates body fat percentage due to disproportionate short stature [26, 48, 56]. Modified anthropometric measurement techniques as maximal hip and umbilical and neck circumferences along height and weight can better capture body composition[55], while dual x-ray densitometry is likely needed for accurate body fat estimation [48, 55]. Cardiovascular and pulmonary

Table 3 Quality appraisal of the included sources, using the NIH Quality assessment tool for observational cohort and cross-sectional studies, NIH Quality assessment tool for systematic reviews and meta-analyses[a], and AGREE II tool[b]

Author	Limitations	Bias	Quality appraisal
Bailey, J. A. et al. [3]	Retrospective data collection, no control group for comparison of abnormality rates	No obvious bias discussed	Fair
Brust et al. [5]	Include small sample size and no control group for comparison	Selection bias is likely, as participants were recruited from a specialty clinic population. Results may not be generalizable to the broader achondroplasia population	Fair
Hooks, M. A. et al. [30]	Single participant. No controls. Short follow-up period	No obvious biases noted	Fair
Hecht, J. T. et al. [26]	Retrospective data, lack of controls	Selection bias of clinic patients	Fair
Hall, J. et al. [25]	Did not appraise quality of evidence presented at conference	Conference presenters were selected by organizers and may not represent all areas of research	NA
Owen, O. et al. (1990)	Small sample size	No obvious biases noted	Fair
Low, L. J. et al. [39]	Search methodology not documented	Quality of the individual studies was not assessed	Fair*
Apajasalo et al. [2]	Lack of details on recruitment and sampling	Possible selection bias as patients were recruited from a national registry. Results may not represent all individuals with skeletal dysplasias	Fair
Mohamed et al. (1998)	Potential survivor bias in oldest age group, self-reported diagnosis of achondroplasia	Selection bias — participants were members of support group Little People of America	Good
Gollust et al. [23]	Potential for selection, information, and confounding biases, the cross-sectional design, and the limited scope of the quality-of-life assessment used	Self-reported survey data, which can be subject to recall bias. Possible selection bias as participants recruited from patient organization membership lists. Results may not be generalizable to the broader population with achondroplasia. Only about 20% of individuals contacted participated	Fair
Carneiro et al. [6]	Single patient, no controls	Reporting bias	Poor
Cervan et al. [9]	Small sample size	Convenience sampling method. Results may not generalize	Fair
Thompson et al. [61]	Quality of original studies in not assessed. Not describe the search strategy or databases used. No details provided on how studies were selected for inclusion. No quality or risk of bias of included studies	Potential selection bias in literature chosen for review. Does not provide details on search strategy and criteria for article selection	Poor[a]
Cortinovis et al. [12]	Small sample size	Convenience sampling, small sample limits generalizability	Fair
Dlugash, R. et al. [16]	Small sample size. Insufficient information in abstract to adequately assess study methodology and quality	Not discussed	Poor
Castro et al. [8]	Very small sample size ($N = 2$), lacks statistical analysis	No obvious biases noted	Fair
Henry, B. et al. [27]	Insufficient information in abstract to adequately assess study methodology and quality	Not discussed	Poor
Henry, B. et al. [28]	Small convenience sample. Self-reported intake	Possible reporting bias	Fair
Schulze, K. et al. [55]	Used DEXA but small convenience sample of only achondroplasia patients	Selection bias	Fair
Schulze, K. J. et al. [56]	Expert discussion without systematic review	Author opinions and perspectives.	Poor
Chiriti et al. [10]	$N = 1$, no control, limited detail on methods and results	Not discussed	Poor
Hoobs D. (2014)	Not a systematic review, so relevant studies may have been missed	No obvious biases noted	Poor[a]
DSAUK [17]	NA	No obvious biases noted	NA

Table 3 (continued)

Author	Limitations	Bias	Quality appraisal
Rodriguez-Gomez, J. et al. [51]	Small convenience sample	Selection bias as participants were recruited from patient association. Results may not generalize	Fair
Rohenkohl et al. [52]	Cross-sectional design limits causal inference. Single sample from a patient organization may not generalize. All measures are self-report. Lack of age-matched general population norms for Germany	Not discussed	Fair
Dhiman, N. et al. [15]	Self-reported survey data	Selection bias as participants recruited from patient organization	Fair
Etayo, L. G. [18]	Opinion piece without systematic methods	Author bias and opinions	Poor
Gomez, E. L. L. et al. [24]	Small convenience sample. Limited pilot testing of prototypes	Not discussed	Good
Da Rocha, L. W., D. et al. (2018)	Very small sample size	Small convenience sample limits generalizability	Fair
Matsushita, M. et al. [43]	Relied only on SF-36, may miss aspects of psychosocial QoL	Selection bias as recruited from patient organizations and hospital clinics. Self-reported survey data subject to recall bias	Fair
Sims, D. T. et al. [60]	Used valid fitness assessments but small sample size. Lacked power for some comparisons	Selection bias	Fair
Jennings, S. E. et al. [35]	Self-reported data, uncertain generalizability	Possible volunteer bias, mostly female/Caucasian sample	Fair
Madsen et al. [40]	Small sample size. Cross-sectional design limits ability to infer causality	Selection bias — recruited from one study	Good
Matsushita, M. et al. [44]	Unclear sampling strategy. Provides insights into patient perspectives but self-reported data prone to bias	Selection bias, reporting bias	Fair
Sims, D. et al. [57]	Small sample size but reasonable given rare condition. Compared to reference data	Selection bias	Good
Sims, D. T. et al. [58]	Lack of imaging to validate marker placement. Sample does not necessarily represent other cohorts with achondroplasia, such as females, the elderly, children, or inactive males	Convenience sampling.	Good
Gattlieb, S. et al. [22]	Snowball sampling limits generalizability. Self-report measures prone to biases	Possible social desirability bias in self-reports	Good
Sims, D. T. et al. [59]	Sample size justification provided. Marker placement limitations acknowledged	Convenience sampling	Good
Constantinides, C. et al. [11]	Did not formally assess risk of bias of included studies. Did not conduct meta-analysis to quantitatively summarize results. Included studies were quite heterogeneous	Publication bias possible as did not search gray literature	Good[a]
de Vries, O. M. et al. [14]	Small sample size. Limited statistical analysis	Selection bias due recruited from one study	Good
Fredwall, S. O. et al. [20]	Small sample size, age differences between groups	Confounding due to age differences between cases and controls	Good
Hoover-Fong et al. [31]	Did not systematically search/identify all studies on topic. Did not formally assess quality of included studies. Perspectives subjective based on experiences of authors	Views of authors may not represent all experts in the field	Good
Ireland, P. J. et al. [33]	Relatively small sample size, limited generalizability	Selection bias due convenience sample recruited through conferences/clinics	Good
Maghnie, M. et al. [41]	Limited to a few European countries, convenience sample	Selection bias due to convenience sample recruited through clinics	Good
Munoz-Reyes, F. et al. [45]	Standard anthropometric measures used but limited details provided	No details provided on sampling	Poor

Table 3 (continued)

Author	Limitations	Bias	Quality appraisal
Rincón Rueda, Z. R. et al. [50]	No validated aerobic and BMI measures, short follow-up period	No obvious biases noted	Fair
Yonko, E. A. et al. [63]	Small sample size. Single clinic population	Selection bias due to single clinic population	Fair
Ajimi, A. et al. [1]	Possible selection and reporting biases, generalizability concerns	Open-ended responses provide insights but prone to reporting bias. Unclear sampling strategy	Fair
Jacinto, M. et al. [34]	No formal risk-of-bias assessment was performed for included studies	No obvious biases noted	Good[a]
Savarirayan, R. et al. [54]	The statements reflect expert opinion more than empirical evidence. Several authors have relationships with industry including BioMarin	The consensus statements highlight the importance of physical activity and ongoing functional assessments at all ages in achondroplasia	5[b]

NA Not applicable

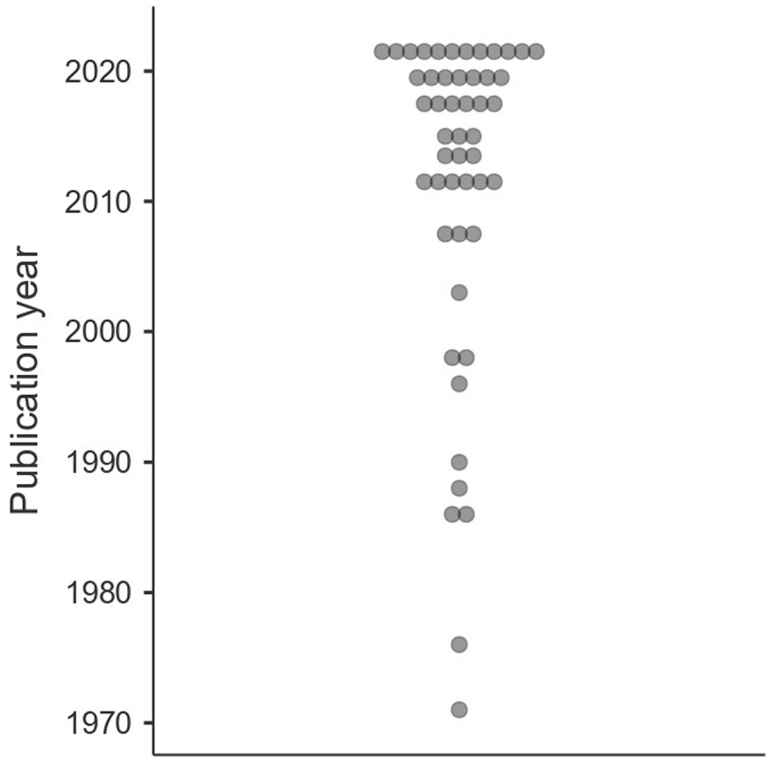

Fig. 2 Distribution of the publication year of the 50 selected records, between 1971 and 2022

responses are impaired during exercise due to anatomic factors. The smaller thoracic cavity and rib cage deformities pose limitations in lung expansion and oxygen intake [37]. A potential chronotropic incompetence constrains heart rate increase, and the disproportionate shorter limbs increase systemic vascular resistance, limiting blood flow and oxygen demands for active muscles [37]. After age 30, physical functioning declines compared to the general population [9] and is further impaired by obesity and muscle weakness, which constrain mobility [10, 30]. Adults with a history of spine surgery also report more physical limitations [1, 30, 44].

Clinical implications and exercise prescription

There is a gap of knowledge on physical fitness assessments validated for adults with achondroplasia, adjusted instruments, and limited reference values [14]. Therefore, for adjusted physical evaluations and cardiorespiratory exercise prescription, the disproportionate short stature, as well the smaller hands and short fingers, should be considered [34]. Also, neurological evaluation to assess grade of lordosis and spinal stenosis [21, 54] should be recommend prior exercise prescription. Adapted sports are a source of multiple benefits, mainly for emotional and social aspects, as they are inclusive and contribute to the overall development of a person with a achondroplasia [50]. Therefore, while being essential to consider the musculoskeletal alterations and potential related health complications, healthcare professionals can introduce and foster opportunities to explore physical activity, exercise, and adapted sports across life span, as a non-pharmacological approach to physical and mental health well-being.

Biomechanical factors

Gait patterns are altered in adults with achondroplasia [57, 58]. Quantitative gait analysis shows impairments in the stride that has a shorter length and higher frequency, along an increased flexion of the ankle and knee and hip internal rotation [31, 50, 59]. When gait is impaired by health complications as secondary osteoarthritis of the hip, it can be improved with physical therapy (Chiriti et al. [10]). When normalized for leg length, stride length is longer, but the frequency is similar compared to average stature adults [59]. Oxygen consumption is also higher due to increased stride frequency, while metabolic cost is higher due to anthropometric differences compared to average stature adults [57, 58, 60]. From studies using the Gait Profile Score, gait deviations were significantly higher in adults with achondroplasia compared to controls [57, 58]. The flexed limb positions are likely a compensation to avoid toe dragging during swing

Table 4 Main conclusions and physical activity-associated factors

Author	Main general conclusions	Factors related to physical activity and exercise
Bailey, J. A. et al. [3]	Elbow radiographic abnormalities are highly prevalent in achondroplasia	The high rate of elbow joint abnormalities found may contribute to upper extremity functional limitations and impact physical activity in achondroplasia
Brust et al. [5]	The study found patients showed good psychiatric adjustment and identity as "little people" despite social challenges. Men tended to have more emotional distress	The study suggested male individuals tended to have more emotional distress, which could negatively impact physical activity
Hooks, M. A. et al. [30]	Indirect calorimetry is more accurate than predictive equations for determining needs in achondroplasia	Unique physiology in achondroplasia may require individual assessment of nutritional needs and be adjusted to activity
Hecht, J. T. et al. [26]	Obesity is common in achondroplasia, starts in childhood, and is 3–8 times more prevalent than general population	High obesity prevalence likely contributes to complications and physical limitations that can negatively impact physical activity capacity
Hall, J. et al. [25]	Multiples discussions between medical experts and families to advance understanding of achondroplasia	The conference highlighted the need for further research on medical and psychosocial factors, which affect physical activity in achondroplasia
Owen, O. et al. [48]	Adults with achondroplasia have lower energy expenditure but higher adiposity compared to general population controls	Lower energy needs may impact nutrition patterns and obesity, which could negatively influence physical activity engagement
Low, L. J. et al. [39]	Anatomical variations inherent in short stature present challenges for physical activity	Individuals with short stature require modifications and adapted programming for successful physical activity participation
Apajasalo et al. [2]	Adults with skeletal dysplasias had lower health-related QoL compared to controls, especially in domains as mobility and pain	Lower mobility and pain can constrain physical activity participation
Mohamed et al. [42]	The functional health status of adults with achondroplasia is not drastically reduced compared to the general US population	Physical activity may be more impaired after 40 years as SF-36. Physical component scores were significantly lower likely due to increased musculoskeletal complaints
Gollust et al. [23]	Adults with achondroplasia had lower quality of life compared to relatives, citing disadvantages related to social barriers as much as health/functioning issues	Social barriers related to achondroplasia may deter physical activity participation to the same extent as health/functioning limitations
Carneiro et al. [6]	Bariatric surgery enabled dramatic weight loss and improved mobility and quality of life in this patient	Provides concrete evidence that obesity-related mobility limitations can be improved with weight loss in achondroplasia, which could enable greater physical activity participation
Cervan et al. [9]	The study found no overall quality-of-life differences between achondroplasia and controls, but achondroplasic women reported less satisfaction with physical and psychological domains	The lower physical domain quality of life score in females with achondroplasia suggests they may experience limitations in physical functioning and activities
Thompson et al. [61]	The review concluded there are serious gaps in research on health, medical, and social factors for adults with skeletal dysplasias. More rigorous research is needed in these areas	The review concluded there are gaps in research on health and social factors that impact physical activity for adults with skeletal dysplasias
Cortinovis et al. [12]	The study highlighted work as a key resource for wellbeing. Social barriers limit social interaction opportunities. Family support was important, but building their own family was a major future goal	The study suggested social barriers remain that could limit physical activity opportunities for adults with achondroplasia
Dlugash, R. et al. [16]	Preliminary results suggest an energy imbalance favoring weight loss, though intake likely underreported	Understanding energy needs and expenditure patterns may help optimize nutrition and exercise recommendations for physical activity in achondroplasia
Castro et al. [8]	Gait abnormalities were found in the subject with achondroplasia compared to the control	Gait and lower limb musculoskeletal abnormalities may impact mobility in achondroplasia. Studies on association with physical activity limitations are needed
Henry, B. et al. [27]	Novel standardized anthropometric methodology established for clinical and research use in adults with achondroplasia	Accurate body composition assessment is important for understanding potential constraints on physical activity in achondroplasia
Henry, B. et al. [28]	Achondroplasia adults have similar macronutrient intake but lower energy/fat/sugar intake compared to average American adults	Nutrition patterns likely influence obesity prevalence and may impact physical activity capacity

Table 4 (continued)

Author	Main general conclusions	Factors related to physical activity and exercise
Schulze, K. et al. [55]	High levels of adiposity along with low muscle mass were found in adults with achondroplasia	Excess fat and reduced lean muscle could negatively impact physical activity capacity
Schulze, K. J. et al. [56]	Customized BMI categorization may be needed in skeletal dysplasias to prevent overdiagnosis of obesity	No concrete evidence, but inaccurate obesity classification could potentially discourage physical activity participation
Chiriti et al. [10]	Clinical and functional indices improved with physical and kinetics treatment	Indicates physical activity and exercise helped reduce pain and improve gait and function in a patient with achondroplasia
Hoobs D. (2014)	Physiological and biomechanical manifestations of achondroplasia impose certain limitations on exercise capacity and training in affected athletes. These require individualized assessment and adapted training programs	Individualized assessment and adapted training programs are needed for athletes with achondroplasia. Certain high-impact exercises may be contraindicated
DSAUK [17]	The examples highlight facilitators and barriers to physical activity participation across the lifespan for individuals with dwarfism/restricted growth	Active involvement in youth sports does not always lead to sustained physical activity in adulthood. Key transition points like start of college or work are often associated with declines in activity
Rodriguez-Gomez, J. et al. [51]	The study suggests the need for preventive interventions and culturally sensitive mental health care among adults with achondroplasia	The high rates of mental health symptoms found could negatively impact motivation and capacity for physical activity
Rohenkohl et al. [52]	Young people with achondroplasia do not differ in generic QoL from norms but report lower condition-specific QoL compared to other short stature diagnoses. Psychological factors like stature attitudes are more associated with QoL than clinical factors	Condition-specific physical QoL is lower compared to other skeletal dysplasias, suggesting greater activity limitations. B. The level of achondroplasia-specific impairment and attitudes are more predictive of QoL than absolute stature. This highlights the potential value of psychosocial interventions
Dhiman, N. et al. [15]	Pain, lack of social support, and perceived stigma were associated with lower quality-of-life scores	The study found pain, lack of social support, and perceived stigma were associated with lower quality of life, which could discourage physical activity
Etayo, L. G. [18]	Nutrition assessment and counselling are important in achondroplasia to address challenges like obesity	Optimizing nutrition may help promote physical activity and fitness
Gomez, E. L. L. et al. [24]	A user-centered design approach generated innovative footwear prototypes and requirements tailored for people with achondroplasia	Adapted footwear may help reduce physical activity barriers and musculoskeletal pain for people with achondroplasia
Da Rocha, L. W., D. et al. (2018)	The study suggested that while prejudice persists, regular physical activity helps promote social inclusion of adults with achondroplasia through improved self-esteem and confidence	Regular physical activity may help promote social inclusion for adults with achondroplasia by improving self-esteem and confidence
Matsushita, M. et al. [43]	The study found reduced physical but not mental quality of life in achondroplasia patients. Care for complications like spinal stenosis is needed to maintain quality of life with aging	Physical quality of life is reduced, and physical activity may be impaired
Sims, D. T. et al. [60]	Adults with achondroplasia use more oxygen during walking but not running compared to average stature adults	Provided direct evidence on higher oxygen cost of walking in achondroplasia. Higher oxygen consumption during walking may make this activity more difficult for achondroplasia
Jennings, S. E. et al. [35]	High prevalence of pain, depression, and anxiety symptoms in skeletal dysplasia population	Mental health issues and pain are common and may negatively impact physical activity
Madsen et al. [40]	High prevalence of abdominal obesity despite low-energy intake. Unhealthy dietary habits may contribute to obesity	Low-energy intake compared to REE despite high obesity prevalence indicates other factors involved. Unhealthy diet may negatively impact health and function. Dietary guidance could help improve outcomes
Matsushita, M. et al. [44]	Adolescents and adults with achondroplasia experience various physical, mental, and social problems that impact quality of life	Musculoskeletal impairments may negatively affect physical activity capacity. Surgery history and aging associated with reduced physical functioning

Table 4 (continued)

Author	Main general conclusions	Factors related to physical activity and exercise
Sims, D. et al. [57]	Adult males with achondroplasia have less total lean and bone mass but higher adiposity compared to average stature males	Studied body composition as it relates to physical activity capacity. Excess fat and reduced muscle mass could impair physical activity capacity
Sims, D. T. et al. [58]	The gait differences are likely due to the need for greater joint flexion to maintain toe clearance during swing phase in achondroplasia	Directly assessed gait biomechanics. Provides insight into gait adaptations that could inform exercise prescription
Gattlieb, S. et al. [22]	Those higher in social dominance orientation showed greater bias against employees with achondroplasia	No concrete evidence on physical activity. Job discrimination could potentially deter people with achondroplasia from sports participation
Sims, D. T. et al. [59]	The increased flexion is likely a biomechanical adaptation to maintain toe clearance during swing phase given the proportionally longer feet	Direct investigation of gait biomechanics. Provides information on gait patterns that could inform exercise prescription
Constantinides, C. et al. [11]	Patients with achondroplasia have poorer outcomes in physical functioning, psychosocial functioning, and quality of life compared to average stature individuals across the lifespan. This appears to be at least partly attributable to disproportionate short stature	Multiple studies show impairments in physical functioning and physical domains of QoL in achondroplasia patients compared to average stature individuals. Psychosocial issues more apparent in adults than children/adolescents. Some evidence that greater height or limb lengthening is associated with improvements in physical functioning and/or physical domains of quality of life
de Vries, O. M. et al. [14]	Adults with ACH have low physical fitness compared to references. 6MWT and 30sSTS are useful	High VO2peak correlation with 6MWT indicates exercise impairment. There is potential for physical activity interventions to improve fitness
Fredwall, S. O. et al. [20]	Despite high BMI, cardiovascular risks appear lower in achondroplasia compared to controls	High BMI in achondroplasia does not appear to confer the same cardiovascular/metabolic risks as in the general population. B. Increased physical activity may be possible without elevated cardiovascular risk in achondroplasia
Hoover-Fong et al. [31]	Although cross-sectional data have described complications of achondroplasia, there is a lack of high-quality longitudinal natural history research across the lifespan. A better understanding of the natural history is critical to optimize patient care and evaluate new therapies	Very little research on physical activity levels or capacity in achondroplasia. Genu varum, spinal stenosis, joint hypermobility, hip flexion contracture, and pain are common issues that may contribute to activity limitations. Lack of longitudinal studies examining interrelationships between physical activity, functional impairments, pain, psychosocial health, and quality of life over the lifespan
Ireland, P. J. et al. [33]	The STEMS is a useful tool to identify mobility aid use and symptoms that impact function across environments in skeletal dysplasia	The STEMS could be used to assess changes in mobility and symptoms in response to interventions aimed at improving physical activity in skeletal dysplasia
Maghnie, M. et al. [41]	Achondroplasia negatively impacts HRQoL across the lifespan	High prevalence of pain negatively impacts HRQoL. B. Associations between short stature and poorer physical HRQoL indicate potential for improvement with therapies to increase height
Munoz-Reyes, F. et al. [45]	Differences in final adult height, weight, and BMI were noted compared to previous studies in other populations with achondroplasia. Possible role of environmental factors on growth variation in achondroplasia	Body composition information that could have implications for weight management and exercise
Rincón Rueda, Z. R. et al. [50]	Physical therapy plays an important role in assessment, diagnosis, and secondary prevention in athletes with achondroplasia	Need for collaboration between physical therapists and coaches to design appropriate training programs for athletes with achondroplasia to optimize performance and minimize injury risk
Yonko, E. A. et al. [63]	Adults with achondroplasia report poor physical and mental QoL. High rates of psychiatric illnesses	Declining physical QoL with age indicates potential for physical activity interventions. The poor physical QoL may be improved with physical activity across lifespan
Ajimi, A. et al. [1]	Adults with achondroplasia/hypochondroplasia experience various physical, mental, and social challenges in daily life. Individual adaptations only addressed some physical/social issues	Musculoskeletal impairments and pain may negatively impact physical activity in ACH/HCH. Individual efforts insufficient to fully overcome limitations

Table 4 (continued)

Author	Main general conclusions	Factors related to physical activity and exercise
Jacinto, M. et al. [34]	There is a lack of research on adapted physical activity and exercise interventions in people with SD. Cross-sectional studies demonstrate this population experiences limitations in physical functioning and quality of life	There are no intervention studies assessing the benefits of adapted physical activity or exercise in SD populations. There are limitations in physical functioning and quality of life where physical activity and exercise interventions may be beneficial
Savarirayan, R. et al. [54]	Functional impairments and activity limitations appear across the lifespan and may be addressed through physical therapy, assistive equipment, and environmental modifications. Pain and obesity should be monitored and managed	Physical activity, strength, and endurance exercise are encouraged as part of healthy lifestyle recommendations

phase due the disproportionately longer feet in relation to leg length, which are flatter, wide, and have a high forefoot pressure indicative of flatfoot [8, 24, 59] which contributed to altered biomechanics [24, 57, 58]. Orthopedic problems in the lower extremities present challenges for physical activity [39] with genu varus (bowing leg deformities) being a common occurrence, caused in part by overgrowth of the proximal fibula [29]. On the upper limbs, repetitive pulling of the triceps on the olecranon apophysis that can cause bone spurs in the elbow [3] is being overall important to consider modifications in exercises and sports requiring high musculoskeletal demands [25, 39].

Psychological and psychosocial factors

Several studies have demonstrated that adults with achondroplasia have substantially lower health-related quality-of-life scores compared to the general population. Reduced physical functioning due to symptoms like chronic pain and obesity appears to be a major contributor, but mental health is also significantly impacted [6, 41, 63]. Over half of adults with achondroplasia have been identified with a psychiatric condition, most often depression or anxiety disorders (Jennings et al. [35]; Yonko et al. [63]). Measured self-esteem is also markedly lower compared to first-degree relatives without the condition [11]. Qualitative research provides additional insight into the psychological struggles faced by many individuals with achondroplasia. Feelings of inferiority, poor self-image, depression, anxiety, and emotional distress are frequently described in relation to the social disadvantages and stigma arising from short stature [23, 51, 61]. Interpersonal challenges are commonly reported due to limited understanding by others about their condition, with over 60% feel stigmatized or treated differently, including by healthcare professionals [1, 15]. Certain periods of life appear to be particularly difficult from a psychosocial perspective [2]. Adolescence and young adulthood can involve social isolation and withdrawal as acceptance from peers declines [2, 61]. However, participation in adapted sports and other social activities can provide psychological benefits by improving self-esteem and confidence (Rocha L., [13]) as well as developing adaptative coping skills and positive attitudes towards short stature [5, 52]. Further research is warranted to understand positive psychosocial experiences like well-being and growth potential in this population (Cortinovis et al. [12]) .

Nutritional factors

Nutritional intake in achondroplasia is an important consideration given the high prevalence of obesity and associated health risks in this population (Madsen et al. [40]; Saint-Laurent et al. [53]). However, the available data on actual dietary intake and energy needs is quite limited. A few small studies have suggested that adults with achondroplasia may consume fewer calories than average compared to the general population [16, 27, 28]. Yet obesity rates remain high, implying a possible imbalance between energy intake, expenditure, and needs. One study found that adults with achondroplasia had lower total 24-h energy expenditure compared to controls, even when adjusted for body composition [48]. Their short stature means stride length and daily energy costs for activities like walking are greater. However, overall metabolic rate may be reduced compared to individuals of average stature [48]. Early dietary and nutritional assessment, nutritional counselling, weight management, and exercise promotion have been emphasized as critical for supporting quality of life across the lifespan [18].

Facilitators of physical activity, exercise, and adapted sports

Several factors may facilitate physical activity engagement among adults with achondroplasia. Improving bone health, body composition, muscle quality, and biomechanics through targeted exercise could enhance physical functioning and make physical activity easier [57, 58]. Understanding specific gait adaptations can inform customized exercise prescription. Also, providing quantitative metrics to track progress may further motivate participation [57, 58]. Developing accurate body mass classification guidelines tailored to short stature could prevent inaccurate obesity labelling that may discourage activity [22]. Access to adapted equipment and modified activities can remove participation barriers. Supportive environments with motivated and knowledgeable coaches and instructors also can foster sports participation, as seen in school and disability sport programs [17]. And focused physical training plans supported by physical therapy can improve joint health, pain, mobility, and fitness [50].

Constraints of physical activity, exercise, and adapted sports

Adults with achondroplasia face numerous constraints that hinder physical activity, exercise, and sports. The altered body composition, low bone mineral density, disproportionate limb ratios, and potential muscle weakness may negatively impact exercise capacity [57, 58]. High rates of obesity, sedentary behavior, and chronic pain further limit activity tolerance [31]. The flexed joint positions and restricted joint range of motion used during gait make certain exercises more difficult, and the short limb length restricts activities that require reach or stride length [59]. Spinal abnormalities and other anatomical

deformities increase injury risks and discomfort [29]. As musculoskeletal issues accumulate with age, mobility decreases, and neurological complications potentially worsen, reducing enthusiasm and capacity for sports [43]. The lack of adapted assessment tools also hampers exercise testing and programming [50]. Accessibility challenges, inadequate adapted equipment availability, as finding appropriate athletic shoes poses a barrier for exercise and sports participation [24]. Also, stigma and waning social support for activity in adulthood were noted as participation barriers 17].

Involvement and participation advocacy organizations in research

On the context on patient centeredness in the evidence base, the level of involvement and participation from advocacy organizations and representatives was limited in the research identified by this scoping review. One relevant example of participation is the research by [24], which utilized a participatory research approach and captured users' perspectives. Regarding the development of this scoping review, we held a focus group with adults with achondroplasia to inform the protocol. Also, one co-author of this scoping review is an adult with achondroplasia and member of ANDO Portugal, the national association for skeletal dysplasias, who provided critical input on the scoping review planning and content.

Strengths and limitations

This scoping review had several strengths, including the use of a protocol to guide the review process, a comprehensive search strategy developed by an experienced librarian, and screening by two independent reviewers. However, some information may be missed at the screening stage. Existent scientific evidence in adults with achondroplasia at multiple levels is quite scarce, and this may compose a bias of interpretation in the conclusions of this review. The included studies were quite heterogeneous in quality, topics, and methods. The quality of many included sources was rated as poor or fair mostly due to small sample sizes, lack of control groups, and reliance on self-reported data prone to recall bias. Selection bias was also a remark, with most studies recruiting participants from advocacy organizations or specialty clinics limiting generalizability. The age factor, with heterogenous age ranges of participants in included references, can pose a limitation, as young adults' perspectives and experiences on physical aspects, biomechanical parameters, and nutritional habits may vary broadly from older adults. These limitations impact the strength of conclusions that can be drawn and highlight the need for more rigorous controlled studies with larger, representative samples, objective outcome measures, and longitudinal follow-up. Also, as this literature search was conducted in October 2022, we acknowledge this as a limitation as more recent evidence may have been published in between.

Conclusions

The existing research on physical activity in adults with achondroplasia as on the factors that impact exercise capacity is still limited, but some interesting findings emerge across the few small studies and sources available. The distinctive anatomical and physiological characteristics of adults with achondroplasia appear to present multifaceted constraints on physical activity engagement across physiological, biomechanical, psychosocial, and nutritional domains. However, substantial knowledge gaps remain. High-quality studies are critically needed to better characterize physical activity capacity, motivators, appropriate programming, and potential benefits across the lifespan for this population. Randomized controlled trials examining the impacts of tailored exercise interventions on outcomes like fitness, pain, function, quality of life, and participation would provide high-level evidence. Also, larger prospective cohort studies correlating long-term physical activity patterns with musculoskeletal health outcomes and functional status are needed to better characterize physical activity capacity, appropriate programming, participation rates, and potential quality-of-life benefits across the lifespan. Implementation studies on optimized adapted sports programs and exercise training models would provide translatable evidence which could ultimately help to improve joint health, pain, muscle strength, and mobility and maintain functionality. And the promotion of regular exercise could be recommended as a non-pharmacologic approach to balance health status and physical fitness aligned with adapted equipment and modified activities in motivating environments. Quantifying biomechanical patterns and fitness metrics can inform personalized exercise prescription, and a multidisciplinary approach including physical therapy may help maintain functionality and improve physical fitness across adulthood. Therefore, a combination of customized strength training, balance programs, targeted exercise interventions, optimized nutrition, and use of population-specific anthropometric assessments and biomechanical models could help address the multidimensional constraints on physical activity participation in achondroplasia.

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Authors' contributions
Conceptualization, IA; methodology, IA and FK; validation, FK, MAC, and CL; formal analysis, IA; investigation, IA; resources, FK and FR; data curation, IA; writing—original draft preparation, IA; writing—review and editing, FK, CL, CDP, ST, OF, and FR; visualization, IA; supervision, CDP, ST, MAC, OF, and F.R; and funding acquisition, FR. All authors have read and agreed to the published version of the manuscript.

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Availability of data and materials
No data has been generated in this review. All information is available in tables presented along the main text.

Declarations

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Not applicable

Consent for publication
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Competing interests
The authors declare that they have no competing interests.

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