SUPPLEMENTARY MATERIAL

TITLE

Considerations for Anthropometry Specific to People with Disproportionate Short Stature

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Table S1 Example data collection sheet.

Patient details		
Date of birth		**Units**
Gestational age (use to correct age for preterm births)		
Corrected age		(Correct age for preterm birth if applicable)
Sex		
Genetic variant		
Parental heights	**Measurement**	**Units**
Mother's height		
Father's height		

Measurement details	Perform measurements according to your local standard operating procedure and input into the table below.					
Visit date						
Age at visit date	**Birth**					
	Average measurement	**Units**	**Average measurement**	**Units**	**Average measurement**	**Units**
Weight						
Height/length						
Sitting height/crown-rump length						
Head circumference						
Arm span						
Upper arm length						
Upper arm circumference						
Waist circumference						
Hip circumference						
Neck circumference						
Notes						

Measurement details	Perform measurements according to your local standard operating procedure and input into the table below.					
Visit date						
Age at visit date						
	Average measurement	**Units**	**Average measurement**	**Units**	**Average measurement**	**Units**
Weight						
Height/length						
Sitting height/crown-rump length						
Head circumference						
Arm span						
Upper arm length						
Upper arm circumference						
Waist circumference						
Hip circumference						
Neck circumference						
Notes						

Table S2 Growth curve resources for children with disproportionate short stature.

Disease or condition	Region or Country	Curve type(s)	Reference	Link(s)
Achondroplasia	Argentina	Height for age Weight for age Head circumference for age	del Pino M, Fano V, Lejarraga H. Growth references for height, weight, and head circumference for Argentine children with achondroplasia. Eur J Pediatr. 2011;170:453–9.	https://doi.org/10.1007/s00431-010-1302-8 (restricted access)
Achondroplasia	Argentina	Sitting height for age Leg length age Sitting height:leg length Sitting height:height Head circumference for age Head circumference: height	del Pino M, Ramos Mejía R, Fano V. Leg length, sitting height, and body proportions references for achondroplasia: New tools for monitoring growth. Am J Med Genet A. 2018;176(4):896–906.	https://doi.org/10.1002/ajmg.a.38633 (restricted access)
Achondroplasia	Australia	Height for age Weight for age Head circumference for age BMI for age	Tofts L, Das S, Collins F, et al. Growth charts for Australian children with achondroplasia. Am J Med Genet A. 2017;173:2189–200.	https://doi.org/10.1002/ajmg.a.38312 (restricted access)
Achondroplasia	China	Height for age	Dai WQ, Gu XF, Yu YG. Exploring the clinical genetic characteristics and height for age growth curve of 210 patients with achondroplasia in China [Chinese]. Zhonghua er ke za zhi (Chinese J Pediatr). 2020; 58(6):461–67.	https://doi.org/10.3760/cma.j.cn112140-20200217-00096 (restricted access)

Disease or condition	Region or Country	Curve type(s)	Reference	Link(s)
Achondroplasia	Egypt	Height for age Weight for age Head circumference for age	Growth charts for Egyptian children with achondroplasia. JCDR. 2019; 13(5):GC01–GC05.	https://www.jcdr.net/articles/PDF/12837/39555_F(KM)_PF1(AJ_SHU)_PFA(AJ_SL)_(NG_SHU)_(AJ_SHU)_PN(SL).pdf (Open Access)
Achondroplasia	Europe	Height for age Weight for age Head circumference for age BMI for age	Merker A, Neumeyer L, Hertel NT, et al. Growth in achondroplasia: development of height, weight, head circumference, and body mass index in a European cohort. Am J Med Genet A. 2018;176:1723–34.	https://doi.org/10.1002/ajmg.a.38853 (restricted access)
Achondroplasia	Europe	Sitting height for age Leg length for age Arm span for age Foot length for age	Merker A, Neumeyer L, Hertel NT, et al. Development of body proportions in achondroplasia: sitting height, leg length, arm span, and foot length. Am J Med Genet A. 2018;176:1819–29.	https://doi.org/10.1002/ajmg.a.40356 (restricted access)
Achondroplasia	Japan	Height for age	Tachibana K, Suwa S, Nishiyama S, et al. A study on the height of children with achondroplasia based on a nationwide survey [in Japanese]. Shounikashinryo (J Pediatr Pract) 1997; 60:1363–69.	n/a
Achondroplasia	Korea	Height for age Weight for age Head circumference for age (up to 3 years)	Lee JS, Shim Y, Cho TJ, et al. Growth patterns of young achondroplasia patients in Korea and predictability of neurosurgical procedures.	https://doi.org/10.1186/s13023-023-02929-6 (Open Access)

Disease or condition	Region or Country	Curve type(s)	Reference	Link(s)
			Orphanet J Rare Dis. 2023;18(1):311.	
Achondroplasia	USA	Length/height for age	Hoover-Fong J, McGready J, Schulze K, et al. A height-for-age growth reference for children with achondroplasia: expanded applications and comparison with original reference data. Am J Med Genet A. 2017;173:1226–30.	https://doi.org/10.1002/ajmg.a.38150 (restricted access)
Achondroplasia	USA	Height for age Weight for age Weight for height Head circumference for age	Hoover-Fong JE, Schulze KJ, Alade AY, et al. Growth in achondroplasia including stature, weight, weight-for-height and head circumference from CLARITY: achondroplasia natural history study-a multi-center retrospective cohort study of achondroplasia in the US. Orphanet J Rare Dis. 2021;16(1):522.	https://doi.org/10.1186/s13023-021-02141-4 (Open Access)
Cartilage-hair hypoplasia	Finland	Height for age Weight for height	Mäkitie O, Perheentupa J, Kaitila I. Growth in cartilage-hair hypoplasia. Pediatr Res. 1992;31(2):176-80.	https://doi.org/10.1203/00006450-199202000-00018 (Open Access)
Diastrophic dysplasia	Finland	Height for age Weight for age	Mäkitie O, Kaitila I. Growth in diastrophic dysplasia. J Pediatr. 1997;130(4):641-6.	https://doi.org/10.1016/s0022-3476(97)70250-6 (restricted access)
Diastrophic dysplasia, spondyloepiphyseal dysplasia congenita, and pseudoachondroplasia	USA	Height for age	Horton WA, Hall JG, Scott CI, Pyeritz RE, Rimoin DL. Growth curves for height for diastrophic dysplasia,	https://doi.org/10.1001/archpedi.1982.03970400034010 (restricted access)

Disease or condition	Region or Country	Curve type(s)	Reference	Link(s)
			spondyloepiphyseal dysplasia congenita, and pseudoachondroplasia. Am J Dis Child. 1982;136(4):316–9.	
Typical growth, with diagnostic cut-offs for hypochondroplasia and Marfan syndrome	Netherlands	Sitting height for age Leg length for age Sitting height:standing height for age	Fredriks AM, van Buuren S, van Heel WJ, et al. Nationwide age references for sitting height, leg length, and sitting height/height ratio, and their diagnostic value for disproportionate growth disorders. Arch Dis Child. 2005;90(8):807–12.	https://doi.org/10.1136/adc.2004.050799 (Open Access)
Hypochondroplasia	Argentina	Height for age Leg length for age Body proportions for age Sitting height:height Head circumference: height ratio	Arenas MA, del Pino M, Fano V. FGFR3-related hypochondroplasia: longitudinal growth in 57 children with the p.Asn540Lys mutation. J Pediatr Endocrinol Metab. 2018;31(11):1279–84.	https://doi.org/10.1515/jpem-2018-0046 (restricted access)
Hypochondroplasia	Europe	Height for age Weight for age Head circumference for age	Cheung MS, Cole TJ, Arundel P, et al. Growth reference charts for children with hypochondroplasia. Am J Med Genet A. 2024;194(2):243–52.	https://doi.org/10.1002/ajmg.a.63431 (Open Access)
Morquio A disease	International	Height for age Weight for age BMI for age	Montaño AM, Tomatsu S, Brusius A, et al. Growth charts for patients affected with Morquio A disease. Am J	https://doi.org/10.1002/ajmg.a.32281 (Open Access)

Disease or condition	Region or Country	Curve type(s)	Reference	Link(s)
			Med Genet Part A. 2008;146A:1286–95.	
Osteogenesis imperfecta	Canada	Height for age	Robinson ME, Rauch D, Glorieux FH, et al. Standardized growth charts for children with osteogenesis imperfecta. Pediatr Res. 2023;94(3):1075–82.	https://doi.org/10.1038/s41390-023-02550-0 (restricted access)
Osteogenesis imperfecta	USA	Height for age	Barber LA, Abbott C, Nakhate V, et al. Longitudinal growth curves for children with classical osteogenesis imperfecta (types III and IV) caused by structural pathogenic variants in type I collagen. Genet Med. 2019;21(5):1233–1239.	https://10.1038/s41436-018-0307-y (Open Access)
Rhizomelic chondrodysplasia punctata	USA	Length for age Weight for age Head circumference for age	Duker AL, Niiler T, Eldridge G, Brereton NH, Braverman NE, Bober MB. Growth charts for individuals with rhizomelic chondrodysplasia punctata. Am J Med Genet A. 2017;173(1):108–13.	https://doi.org/10.1002/ajmg.a.37961 (restricted access to full manuscript, but curves available for download as Supplementary figures)
Spondyloepiphyseal dysplasia congenita and other COL2A1 related dysplasias	International	Height for age	Terhal PA, van Dommelen P, Le Merrer M, et al. Mutation-based growth charts for SEDC and other COL2A1 related dysplasias. Am J Med Genet C Semin Med Genet. 2012;160C(3):205–16.	https://doi.org/10.1002/ajmg.c.31332 (restricted access)

Abbreviations: BMI = body mass index; COL2A1 = collagen, type II, alpha 1; USA = United States of America.

Table S3 Additional anthropometric measurements to consider for people with disproportionate short stature.

Measurement	Notes on technique	When to consider
Lower arm (forearm) length	Measure from the olecranon process of the elbow to the distal lateral process of the ulna, parallel to the lateral surface of the forearm.	In conditions in which distal limb segments typically exhibit disproportion.
Hand: Palm and middle finger length	Patient should be sitting with arm bent at elbow and palm facing up. Palm: Measure from the distal crease of the wrist to the proximal flexion crease of the middle finger. Middle finger: Measure from the proximal flexion crease of the middle finger to the tip of the middle finger.	In conditions in which hands exhibit disproportion or anatomical differences.
Hand: Palm width	Measure at the level of the metacarpophalangeal joints, from the edge of the hand on one side, across the palm, to the edge of the hand on the other side.	In conditions in which hands exhibit disproportion or anatomical differences.
Full leg length (spina-malleolar distance)	Patient should be lying down. On each leg, measure from the anterior superior iliac spine to the medial malleolus.	In conditions in which unequal leg lengths cause a difference in the height of the iliac crest in the upright position. This measurement is approximately the same as the distance from the top of symphysis pubis to the floor and can therefore be used to estimate the lower body segment length for patients who cannot keep an upright position.
Upper leg (thigh) length	Patient should be seated. Measure along the anterior aspect of the leg (‘top’ while in seated position), from the midpoint of the inguinal crease to the proximal edge of the patella.	In conditions in which proximal limb segments typically exhibit disproportion.

Measurement	Notes on technique	When to consider
Knee to foot height	This is best measured with a knee height caliper (segmometer is an alternative). The patient should sit upright on a stool with knee and ankle both at 90° angles and legs hanging freely. Place one end of the caliper under the heel of the foot directly below the ankle bone, the other end is placed on the anterior surface of the thigh approximately above the head of the fibula. The caliper shaft should align with the long axis of the leg. Apply slight pressure for an accurate measurement.	In conditions with disproportion in limb segments, and in patients with kyphosis or scoliosis.
Tibial length	This is best measured with a Campbell caliper (segmometer is an alternative). One end is placed at the medial epicondyle of the femur and the other end at the distal tip of the medial malleolus (ankle bone). The caliper shaft should be parallel to the long axis of the leg. Apply slight pressure for an accurate measurement.	In conditions with disproportion in limb segments, and in patients with kyphosis or scoliosis.