

# Developmental Milestones in Infants and Young Australasian Children With Achondroplasia

Penelope Jane Ireland, BPhy,\* Sarah Johnson, BSpPath,\* Samantha Donaghey, BOccthy,\* Leanne Johnston, PhD,† James McGill, MD,‡ Andreas Zankl, MD,§ Robert S. Ware, PhD,¶¶ Verity Pacey, BPhy,\*\* Jenny Ault, MBBS,†† Ravi Savarirayan, MD,‡‡ David Sillence, MD,§§ Elizabeth Thompson, MD,||| Sharron Townshend¶¶¶

**ABSTRACT:** *Background:* Achondroplasia, the most common form of chondrodysplasia (inherited skeletal dysplasia), is characterized by a significant delay in the development of communication and motor skills, particularly during the first 2 years. Although some information regarding timing of development for children with achondroplasia is available, no study has evaluated simultaneously the pattern of skill development across multiple key developmental areas. *Method:* This study used a retrospective questionnaire to quantify developmental data on milestone achievement. Twenty families of children with achondroplasia throughout Australia and New Zealand were asked to document age of acquisition for 41 gross motor, fine motor, and communication and feeding milestones. More than one half of the items assessed were milestones identified in the Australian State Government Personal Health Record Books. The results are compared with previously available information regarding development of motor skills by a cohort of American children with achondroplasia. *Results:* Although the results support previously reported delays in gross motor and communication skill development, fine motor development does not seem to be as delayed as previously suggested. Information on development of self-feeding skills is presented for the first time and occurs later in this group than the typically developing population. We describe 2 distinctive and previously unreported methods of transitioning between static positions commonly used by children with achondroplasia. *Conclusion:* Delays were reported across gross motor and communication and feeding skills but were not observed during development of fine motor skills. Additional information is also offered regarding a variety of unusual movement strategies demonstrated by young children with achondroplasia.

(*J Dev Behav Pediatr* 31:41–47, 2010) **Index terms:** achondroplasia, skill development, Australia, skeletal dysplasia, developmental milestones.

**A**chondroplasia is the most common form of short-limbed short stature, occurring with an incidence of between 0.36 and 0.6 per 10,000 live births.<sup>1</sup> Children

From the \*Queensland Paediatric Rehabilitation Service, Royal Children's Hospital, Brisbane, Queensland, Australia; †Division of Physiotherapy, School of Health and Rehabilitation Sciences, University of Queensland, Brisbane, Queensland, Australia; ‡Department of Metabolic Medicine, Royal Children's Hospital, Brisbane, Queensland, Australia; §University of Queensland Centre for Clinical Research, Royal Brisbane and Women's Hospital, Brisbane, Queensland, Australia; ¶School of Population Health, The University of Queensland, Brisbane, Australia; ¶¶Queensland Children's Medical Research Institute, Queensland, Brisbane, Australia; \*\*Department of Physiotherapy, The Children's Hospital at Westmead, Sydney, New South Wales, Australia; ††Department of Paediatric Rehabilitation, The Children's Hospital at Westmead, Sydney, New South Wales, Australia; ‡‡Victorian Clinical Genetics Service, Royal Children's Hospital, Melbourne, Victoria, Australia; §§Department of Clinical Genetics, The Children's Hospital at Westmead, Sydney, New South Wales, Australia; |||South Australia Clinical Genetics Service, Women's and Children's Hospital, Adelaide, South Australia, Australia; ¶¶Genetics Service of Western Australia, King Edward Memorial Hospital, Perth, Western Australia.

Received June 2, 2009; accepted October 2, 2009.

This study was funded through an Allied Health Research Grant from the Royal Children's Hospital Foundation, Brisbane.

Address for reprints: Penelope J. Ireland, BPhy, Queensland Paediatric Rehabilitation Service, Royal Children's Hospital, Herston Road, Herston 4029, Brisbane, Queensland, Australia; e-mail: penny\_ireland@health.qld.gov.au

Copyright © 2010 Lippincott Williams & Wilkins

with achondroplasia have a number of well-recognized anatomical differences, including a disproportionate short stature, with marked shortening of the limb segments, macrocephaly, generalized hypotonia, generalized joint hypermobility, increased lumbar lordosis, thoracolumbar kyphosis, and reduced elbow extension. The head generally appears to be large with a prominent forehead and chin and a flattened midface region.<sup>2–4</sup> Although older children with achondroplasia are considered within the normal range of cognitive functioning, a number of researchers have identified that children with achondroplasia generally have significantly delayed development of motor and communication skills, particularly during the first 2 years.<sup>4–7</sup> Overall, little information on development is available, especially for communication and feeding skills and no information specific to the Australian and New Zealand populations.

Developmental delays observed in children with achondroplasia were first reported by Todorov et al,<sup>5</sup> who assessed the performance on 15 gross motor and communication skills of 166 adults and 31 American children with achondroplasia, using a retrospective questionnaire based on the Denver Developmental Screening Test.<sup>8</sup> The Denver Developmental Screening

Test was designed to give a brief overview of a child's development, based on the performances of 1036 typically developing children in 1966. A major revision and restandardization of the Denver Developmental Screening Test (Denver II) was published in 1992 and included a review of individual items, inclusion of additional items, and standardization of the tool on a total of 2096 children.<sup>9</sup> In 1997, Fowler et al<sup>7</sup> prospectively monitored 93 children with achondroplasia, reaffirming the motor delays previously reported by Todorov et al<sup>5</sup> and describing a number of specific and unusual preambulation locomotion patterns utilized by children with achondroplasia. These included "snow ploughing" and "reverse snow ploughing" in which the head acts as a pivot point, whereas the legs and feet propel the child along. They also described "spider crawling"; a type of locomotion in which the child supports the weight on hands and feet as opposed to the hands and knees as seen in a more traditional crawling pattern. Fowler et al<sup>7</sup> concluded that the developmental delays seen for gross and fine motor skills, and the unusual forms of preambulant locomotion, are compensatory for the anatomical differences in children with achondroplasia. No information has previously been provided on specific sequences of movement demonstrated by children with achondroplasia when transitioning between positions. Although speech and language delay is well documented for children with achondroplasia,<sup>4,10</sup> and a review by a speech and language pathologist is recommended within the first 2 years,<sup>4</sup> there is little quantitative information concerning when communication milestones are reached. No information concerning the development of feeding skills has been previously reported.

The purpose of our study was to collect detailed information regarding the age and sequence of development across the areas of gross motor, fine motor, and communication and feeding skills for a cohort of Australian and New Zealand children with achondroplasia and to compare these data with that reported in the previous American-based cohorts. A secondary aim was to describe the characteristic sequences of movements demonstrated when children with achondroplasia transition between static positions.

## METHODS

The project was developed as an Australasian population-based study. Participants were recruited through the Bone Dysplasia and Clinical Genetics clinics at major hospitals in all 6 Australian states, through the Short Statured People Association branches in Australia and through the Little People of New Zealand Association. Ethical approval was obtained through the Ethics in Human Research Committees in each state. Informed consent was obtained from parents or guardians using the protocols recommended by the Ethics in Human Research Committees for each state.

Individuals were eligible if they had a medical diagnosis of achondroplasia. From June 2001, we began enrolling all individuals with achondroplasia aged younger than 5 years. On recruitment, participants were asked to recall the age at which their child had achieved listed milestones. The achievement of any milestones not achieved at recruitment was then recorded prospectively until the child turned 5 years. Children younger than 15 months at the time of initial contact were recruited to the prospective component of the study, which is currently under way. Children with additional neurological problems, not related to achondroplasia, such as epilepsy or cerebral palsy, were excluded due to the potential for these conditions to affect developmental progress.

A 41-item written questionnaire was developed using a multidisciplinary reference group of expert clinicians and researchers working with children with achondroplasia. It covered 41 standard milestones across gross motor (14 items), fine motor (8 items), communication (12 items), and feeding (7 items) domains commonly evaluated in clinical practice through parental questioning. Twenty-four of the 41 items were milestones identified in the Australian State Government Personal Health Record Books (developed by the Australian College of General Practitioners, the Australian Medical Association, the Australian College of Paediatrics, the Australian College of Midwives, and the Professional Association of Child Health Nurses). These books are issued to all children born in Australia and New Zealand at the time of their birth and are used by parents and health professionals to monitor a child's development and medical history throughout the first 5 years of their lives. The developed items were cross-referenced against recognized standardized assessments to ensure relevance from a developmental perspective. Thirty-two items covering gross motor, fine motor-adaptive, and language and personal-social skills were reported in the Denver Developmental Screening Test II (26 items) and the Rossetti Infant Toddler Language Scale (6 items).<sup>11</sup> An additional 3 feeding items (looking at progression through puree and mashed consistencies to finger feeding) were developed by an expert multidisciplinary team panel with input from a speech and language pathologist, occupational therapist, physiotherapist, and medical practitioner.

Parents were asked to report on 5-specific forms of preambulant locomotion common in this population including snow ploughing, reverse snow ploughing, commando crawling, "bear" or "spider" walking, and traditional crawling (5 items) based on the study by Fowler et al.<sup>7</sup> Parents were also asked to describe movement sequences utilized by their child when transitioning from lying to sitting, standing to sitting, and sitting to standing (3 items). The test items did not require specific skills to administer and the questionnaire included pictorial representation and a brief description where applicable.

After providing consent, families were sent the questionnaire and asked to record the age of achievement in months and weeks, for each of the 41 items. If parents

were uncertain of the age of achievement, they were asked to record this as “unknown” (although only 5% of total items were recorded in this way). Families were resent the questionnaire at 3 monthly intervals and asked to record their child’s development for the previous 3-month period, until all 41 items were demonstrated and recorded.

## Data Analysis

For each of the 41 milestone items, minimum, maximum, median, and 90th percentile ages for milestone attainment were calculated. All items were achieved by 5 years (60 months). Data were presented in a similar format by Todorov et al<sup>5</sup> after their retrospective assessment of 166 adults and 31 children (no age ranges offered) and Fowler et al<sup>7</sup> who reported on their prospective assessment of 93 children during a 14-year period, from 1 to 60 months (no mean age of recruitment reported). The 90th percentile ages for each of the common items recorded by Todorov et al,<sup>5</sup> Fowler et al,<sup>7</sup> and the Denver II<sup>9</sup> are included for comparison. For the preambulation locomotion strategies and transitional movement sequences, the percentage of respondents demonstrating each movement is presented.

## RESULTS

Twenty-eight children (17 boys) were identified on databases of Bone Dysplasia services or support groups across Australia and New Zealand. Parents of all children were contacted by mail and invited to participate in the study. Twenty (71%) families enrolled and completed all questionnaires. Age at enrollment ranged from 1 child enrolled at 20 months, 7 children enrolling between 26 and 36 months, 6 children between 37 and 48 months, and 6 children between 49 and 60 months, with a mean age of recruitment of 40.6 months. Thirteen participants were boys and 7 were girls.

### Gross Motor Skills

As expected, the average age of acquisition of gross motor milestones for children with achondroplasia was later in all situations when compared with results for typically achieving children as reported by the Denver II (Table 1).

### Movement Strategies

Commando crawling was the most popular method of preorthograde locomotion observed in the Australian and New Zealand cohort, demonstrated by 85% of our sample (Table 2).

Two distinctive and previously unreported methods of transitioning between positions were identified in the Australian and New Zealand cohort (Table 3). When transitioning from lying to sitting, the most common strategy was for the child to lie prone, abduct the legs fully into a “split-like” position, push up through the arms, and extend the head until they were able to lever into a more upright position (demonstrated by 60% of children). When pulling to stand from a lying or sitting

position, 55% of children adopted a “split-like” position in prone or sitting, allowing close access to an object and then used their arms to pull up into a standing position.

### Fine Motor Skills

With regard to time taken to develop fine motor skills, there was little difference between those milestone achievements reported for typically developing children by the Denver II,<sup>9</sup> particularly for the earlier milestones (Table 1).

### Communication Skills

The current Australasian achondroplasia cohort took longer to achieve most communication milestones than the typically developing cohort described by the Denver II<sup>9</sup> (Table 1).

### Feeding Skills

Evaluation of eating and drinking skills in the Australian and New Zealand cohort showed that of the 20 children who participated in the study, 10 (50%) were reported to successfully breastfeed; 9 (45%) children were bottle-fed; and 8 (40%) combined breast and bottle feeding. Infants in Australia are generally introduced to solid food between 4 and 7 months.<sup>12-14</sup> The timing of the introduction of pureed solids for children with achondroplasia was similar to that seen in the general populace. Independent feeding skills attainment occurred at an older age in this population than that reported in the normative population by the Denver II (Table 4).

## DISCUSSION

We have compiled data regarding the age and sequence of development across the areas of gross motor, fine motor, and communication and feeding skills for a cohort of Australian and New Zealand children with achondroplasia, including, for the first time, feeding skill acquisition. The pattern of milestone attainment recorded for this group supports previous reports of delay in gross motor and language skills,<sup>5,7</sup> although fine motor skill development seems to be an area of relative strength for children with achondroplasia, with less delays reported than previously.<sup>7</sup>

Identifying a specific profile of development is necessary for conditions such as achondroplasia, both for guiding future development of specific treatment strategies,<sup>5,15</sup> and where unusual delays in skill attainment or demonstration of varied movement strategies may be seen as indicative of significant developmental delay by inexperienced clinicians.<sup>7</sup> More detailed documentation of milestone attainment, across a variety of skill areas, offers health practitioners a clearer reference point with which to compare a child’s performance and identify those children performing outside the level expected for their age and medical condition-related profile.<sup>16</sup> The provision of more detailed information about the developmental journey in the early years for groups, such as children with achondro-

**Table 1.** Percentile Distribution for Timing of Acquisition of Gross Motor, Fine Motor, and Communication Skills (mo) for Children With Achondroplasia

Milestone	Sample Size	Percentiles (mo)				
		Median (Min–Max)	90th	90th (Todorov)	90th (Fowler)	90th (Denver II)
Gross motor						
Lift head when lying on stomach <sup>a</sup>	17	5 (1.5–10)	6.7	9		3.5
Roll over <sup>a</sup>	18	7 (3–9.5)	9	8	20	5.2
Snow plough	11	14 (6–36)	24			
Reverse snow plough	6	9 (6–11)	10.8			
Commando crawl	17	11 (6–18)	16.2			
Bear walking	10	16 (11–30)	27.3			
Traditional crawling <sup>a</sup>	5	16 (9.5–30)	25.2	13	14.5	
Into sitting from lying <sup>a</sup>	14	15.1 (8–24)	21			10
Into sitting from standing	16	16 (10–42)	26.5			
Into standing from sitting <sup>a</sup>	19	16 (8.5–48)	24.4	18	20	10
Stand holding on <sup>a</sup>	17	13.5 (5–40)	25	21	19	12
Stand unsupported	19	17 (12–43)	35.2	23	29	14
Walk holding on <sup>a</sup>	19	18 (12–48)	36.4	22	27	12.7
Walk independently <sup>a</sup>	20	21.2 (14–52)	38.2	30	22.5	15
Fine motor						
Reach for object <sup>a</sup>	13	5 (2–8)	7		15	5.5
Pass objects <sup>a</sup>	13	7 (4–10)	8.8		14	7.5
Bang objects together	9	7.8 (5–12)	9.6		14	11
Scribble with crayon <sup>a</sup>	14	17 (5–36)	24		30	17
Unscrew lid from jar	13	30 (24–48)	33.2			
Draw circle <sup>a</sup>	13	30 (24–42.5)	36		48	45
Build tower 2 blocks <sup>a</sup>	13	15 (6–40)	30		29.5	20
Build tower 8 blocks <sup>a</sup>	14	24 (7–48)	34.4		35	42
Communication						
Smile <sup>a</sup>	18	2.1 (0.8–6)	5			2
Babble <sup>a</sup>	18	7.3 (3–30)	15	10		9
Wave <sup>a</sup>	16	11 (8–36)	25			14
Say “mama” <sup>a</sup>	16	10.3 (6–24)	20	17		13
Shake head	9	13 (9–25)	19.5			
Peek-a-boo	13	12 (3–36)	19.6			9.7
1-step request <sup>a</sup>	14	18.8 (8.5–36)	29.4			
Identify body parts	15	22 (7.5–40)	26.4			27
Imitate words	13	18 (11–30)	28.8			
Use single words <sup>a</sup>	14	19.9 (10.5–36)	28.8			15
Combine 2 words <sup>a</sup>	16	24 (12–48)	34	24		25
Short sentences <sup>a</sup>	16	27.5 (17–54)	37.6	36		

<sup>a</sup>Denotes those items found in the Australian State Government Personal Health Record Books.

plasia, may assist in making the initial disclosure of diagnosis less distressing for families.<sup>17</sup>

### Gross Motor Skills

Comparisons of gross motor skill development in our study with the previous studies<sup>5,7</sup> identified both similarities and differences existing among the three groups. The

American Academy of Pediatrics and Committee of Genetics developed Health Supervision Guidelines for children with achondroplasia in 1995 (revised and updated in 2005), which guide the clinical management of children with achondroplasia. After the development of these recognized guidelines, there is no reason to expect major differences in overall clinical management, particularly be-

**Table 2.** Preorthograde Movement Strategies Use by Children With Achondroplasia

Movement Strategy	Number/Percentage (n = 20)	Average Age (mo) (Ireland et al, in This Issue)	Number/Percentage <sup>7</sup> (n = 35)	Average Age (mo) <sup>7</sup>
Snow plough	15 (75)	16.6	21 (60)	12.2
Reverse snow plough	8 (40)	8.9	15 (43)	12.2
Commando crawl	17 (85)	11.4	27 (77)	16.6
Bear walking	10 (50)	18.1	8 (23)	17.6
Traditional crawling	5 (25)	17.7	7 (20)	9.6

tween the cohort in the study by Fowler et al<sup>7</sup> and our current cohorts, although variations may exist after taking into account individual patient circumstances including whether management of individuals within the earlier American cohorts involved the specific restrictions to early sitting that are now clearly recommended. This was not discussed in either of the previous American studies. Variations in findings may be attributed to the retrospective collection of data used in this study, the relatively small numbers in the study group when compared with the larger numbers in the American-based studies and possible impact on skill development by possible differences in clinical management between groups, such as restriction of early sitting. However, the similarity in trends for later gross motor milestone achievement supports previously documented reports of delay in gross motor skill development for children with achondroplasia.

Our results reaffirm the findings of Fowler et al,<sup>7</sup> based on a retrospective study of 35 children, that many children with achondroplasia demonstrate unusual preorthograde stages of ambulatory development. Fowler et al<sup>7</sup> concluded that these forms of locomotion should not therefore be considered abnormal but as specific strategies adopted by these children that are adaptive to the biomechanical challenges associated with achondroplasia such as hypotonia, macrocephaly, rhizomelia, and

joint hypermobility. Our current results support the original findings of Fowler et al<sup>7</sup> that children with achondroplasia seek out movement strategies for both locomotion and transitioning between positions where the effects of the biomechanical changes are reduced.

This study presents a number of specific sequences of movement utilized by children with achondroplasia when initially learning to transition between positions. The unusual method of incorporating full hip abduction into some transitional movements would seem to be an adaptive strategy, allowing these children to use their joint hypermobility as a mechanism for reducing the biomechanical impact of the rhizomelia and the macrocephaly.

### Fine Motor Skills

The time frames demonstrated by the Australian and New Zealand cohort for fine motor skill development were more similar to the typically developing population, suggesting that fine motor development may not be as delayed as previously thought. One possible explanation is that the limitations on sitting recommended for infants with achondroplasia to reduce risk of further kyphotic development<sup>6</sup> necessitates that these children spend more time in very stable positions such as supine and prone. A delayed ability to sit independently (mean of 15.1 months) may allow children the opportunity to practice more bilateral and bimanual hand tasks. Although the characteristic brachydactyly and trident configuration of the fingers alters the biomechanical properties of the hands, the increased proximal stability afforded by working in supine and prone positions may help to counteract the influences of the additional biophysical features such as the hypotonia, joint hypermobility, and macrocephaly. Furthermore, because of the limited positions and lack of sitting positions available to these infants during the first 12 months, parents and caregivers may be offering their child greater opportunities to develop and practice in hand manipulation skills, by introduction of a wide variety of fine motor toys and activities. These children may have already developed a number of compensatory strategies to improve their in-hand manipulation skills, before adopting positions such as sitting, where increased postural demands are placed on them. Further research with a larger cohort of children is necessary to further evaluate the development of these skills.

**Table 3.** Transitional Movement Strategies Used by Children With Achondroplasia

Transition	Movement Strategy	Number/Percentage of Children Using Strategy (N = 20)
Into sitting from lying	Roll onto one side	5 (25)
	Push up from stomach	12 (60)
	Others	1 (5)
From standing up into sitting	Dropping backwards	12 (60)
	Squatting down	6 (30)
	Spreading legs	4 (20)
Pull into standing from sitting down	Kneeling up on both knees	6 (30)
	Lying on stomach	11 (55)
	Others	3 (15)

**Table 4.** Percentile Distribution for Timing of Acquisition of Feeding Skills (mo) in Children With Achondroplasia

Milestone	Sample Size	Percentiles (mo)		
		Median (Min–Max)	90th	90th Denver II
Cup drinking <sup>a</sup>	15	20 (7–36)	31.2	18
Puree/smooth solids	18	5 (3–9)	7.3	
Mashed solids	19	5 (5–34)	18.4	
Finger feeding	15	15 (7–30)	24	7
Self-feed with spoon <sup>a</sup>	16	20.5 (9.5–50)	33	20

<sup>a</sup>Denotes those items found in the Australian State Government Personal Health Record Books.

## Communication Skills

Although the children in our group reached each of the communication milestones later than the group studied by Todorov et al,<sup>5</sup> the last reported language acquisition skill (using short sentences) occurred at similar ages in both groups (37.6 months compared with 36 months). This supports previous findings that delays in the acquisition of speech are common in children with achondroplasia. Possible explanations for the delay in acquisition of speech may be related to the frequent occurrence of acute and chronic otitis media within this population group.<sup>10,18</sup> Hunter et al<sup>10</sup> reported that in a multicentre patient review, 35% of 1- to 2-year-old children and 37% of 2- to 3-year-old children received at least one set of grommets or ventilation tubes. The transient hearing loss associated with otitis media may then have a significant influence on speech acquisition.<sup>10</sup>

## Feeding Skills

This study is the first to include an assessment of the timing of feeding skill acquisition for children with achondroplasia. The extension of the age of attainment for feeding skills beyond that reported in the normal population (Denver II) is information that has not been previously identified in children with achondroplasia. One possible reason for the delay in feeding skills seen in this sample may be linked to the biophysical features of achondroplasia. Morris and Klein<sup>19</sup> noted that the efficient use of the mouth for eating relies largely on the steadiness or stability of the trunk, neck, and head. Children with postural difficulties may lack the foundational support and stability necessary for the refined motor skill actions required by an eating task. The delays in developing skills in positions up against gravity experienced by children with achondroplasia, related to challenges such as hypotonia and delayed head control, may also influence the child's ability to use the mouth efficiently. Feeding difficulties may also be linked to central nervous system impairment, e.g., swallowing difficulties can be caused by hydrocephalus or brainstem compression, both of which are can be associated with achondroplasia. Another factor to be considered in any delay of feeding skills is the external influence relating to the delayed sitting for infants with achondroplasia. Placement in a reclined position, necessary to minimize further kyphotic

development in children with achondroplasia,<sup>6</sup> alters the dynamics of chewing and swallowing and may contribute to challenges experienced by infants when moving to more lumpy textures. The delay in developing self-feeding skills may also be associated with the rhizomelic limb shortening and lack of full elbow extension in this population.

There are several limitations to this study that must be considered. Although data were collected from all consenting families from the Australasian cohort, which represented >70% of the identified population of children aged between 15 months and 5 years in Australia and New Zealand, it remains a relatively small sample at 20 children. Because of the nature of our study design, it is possible that we have not contacted all eligible children. Nevertheless, using the identified population to calculate incidence of achondroplasia results in an estimated incidence rate of 0.23 per 10,000 births, a rate similar to that reported by Waller et al,<sup>1</sup> suggesting the majority of children were identified.

The use of retrospective data collection and parental questionnaire is likely to reduce reliability and accuracy of reported information for all but the most easily remembered milestones such as “walking independently,”<sup>15</sup> and may particularly influence recall of timing for communication and fine motor skills. A child's position within the family may also influence the parent's experience of development and reporting ability and should be considered in future studies. To reduce the potential bias associated with recall, more than one half of the items on the questionnaire were also identified in the child's individual Personal Health Record Book, allowing families to utilize information previously recorded through attendance at child health and medical appointments. To examine our data for systematic age-at-reporting bias, we compared data from the 8 children enrolled at 3 years or younger against the 12 children enrolled when aged older than 3 years. We compared median time to achievement: for 49% of items, median age at achievement was less for the group enrolled at 3 years or younger, for 44% of items, median age at achievement was less for the older group, for 7% of items, median age at achievement was the same for both groups. These results suggest there is no consistent pattern of misreporting dependent on enrollment age. Many parents in this retrospective group demonstrated a high level of recall (recording both weeks and months) for each of

the items, with families clearly monitoring common developmental milestones. For conditions where frequency and prevalence are low in the general community, a retrospective analysis remains one of the most practical ways to clinically gather information regarding development. The limitations associated with results of a retrospective study can then only be countered by results from a prospective, longitudinal study.<sup>20</sup> A prospective study looking at gross motor, fine motor feeding, and communication skills is currently under way to more thoroughly understand development in Australasian children with achondroplasia. It is anticipated that data collection for the prospective group will be completed within the next 2 to 3 years.

## CONCLUSION

These findings will prove useful to families and clinicians working with young children with achondroplasia, particularly during the first 2 to 3 years. They support previous reports of both developmental delays and unusual movement strategies in children with achondroplasia. The delays in gross motor and communication skills seem consistent with previously reported results, although there seems to be less influence on the development of fine motor skills than previously thought. Information on feeding milestones has been reported for the first time. The results of this study will add to the currently available knowledge regarding developmental information for children with achondroplasia and assist both families and clinicians to monitor a child's progress. More detailed developmental profiles for this group across a variety of areas will reassure families that their child's development does not differ in their developmental sequence from other children with achondroplasia. Further research, including prospective studies, is necessary to evaluate the timing of development across multiple areas simultaneously and ascertain the development of participation skills within functional tasks such as eating and dressing.

## ACKNOWLEDGMENTS

We thank all the families who generously donated their time for this research. Without their help, this study would not have been possible.

## REFERENCES

1. Waller DK, Correa A, Tuan MV, et al. The population-based prevalence of achondroplasia and thanatophoric dysplasia in selected regions of the US. *Am J Med Genet A*. 2008;146:2385-2389.

2. Nehme AM, Riseborough EJ, Tredwell ST. Skeletal growth and development of the achondroplastic dwarf. *Clin Orthop Relat Res*. 1976;116:8-23.
3. Scott CI. Achondroplastic and hypochondroplastic dwarfism. *Clin Orthop Relat Res*. 1976;114:18-29.
4. Trotter TL, Hall JG; and Committee on Genetics. Health supervision for children with achondroplasia. *Pediatrics*. 2005; 116:771-783.
5. Todorov AB, Scott CI, Warren AE, Leeper JD. Developmental screening tests in achondroplastic children. *Am J Med Genet A*. 1981;9:19-23.
6. Hall JG. Kyphosis in achondroplasia: probably preventable. *J Pediatr*. 1988;112:166-167.
7. Fowler ES, Glinski LP, Reiser CA, Horton VK, Pauli RM. Biophysical basis for delayed and aberrant motor development in young children with achondroplasia. *J Dev Behav Pediatr*. 1997; 18:143-149.
8. Frankenburg WK, Dodds JB. The Denver Developmental Screening Test. *J Pediatr*. 1967;71:181-191.
9. Frankenburg WK, Dodds JB, Archer P, Shapiro H, Bresnick B. The Denver II: a major revision and restandardization of the Denver Developmental Screening Test. *Pediatrics*. 1992;89: 91-97.
10. Hunter AG, Banksier A, Rogers JG, Sillence D, Scott CI Jr. Medical complications of achondroplasia: a multicentre patient review. *J Med Genet*. 1998;35:705-712.
11. Rossetti L. *The Rossetti Infant-Toddler Language Scale*. East Moline, IL: LinguiSystems; 1990.
12. Donath SM, Amir LH. The introduction of breast milk substitutes and solid foods: evidence from the 1995 National Health Survey. *Aust NZ J Public Health*. 2002;26:481-484.
13. Boulton J, Landers M. *The Toowoomba Children's Nutrition Study 1993-1997*. Toowoomba: Darling Downs Public Health Unit; 1999.
14. Graham VA, Gibbons K, Marraffa C, Henry L, Myers J. Filling the gap: weaning practices of children aged 0-2 years in western Metropolitan Melbourne. *J Paediatr Child Health*. 1998;34:513-517.
15. Engelbert RH, Uiterwall CS, Gulmans VA, Pruijs HE, Helders PJ. Osteogenesis imperfecta: profiles of motor development as assessed by a postal questionnaire. *Eur J Pediatr*. 2000;159:615-620.
16. Crockett MM, Carten MF, Hurko O, Sponseller PD. Motor milestones in children with diastrophic dysplasia. *J Pediatr Orthop*. 2000;20:437-441.
17. Hill V, Sahhar M, Aitken M, Savarirayan R, Metcalfe S. Experiences at the time of diagnosis of parents who have a child with a bone dysplasia resulting in short stature. *Am J Med Genet A*. 2003;122:100-107.
18. Cohen MM. Dysmorphic syndromes with craniofacial manifestations. In: Stewart RE, Prescott G, eds. *Oro Facial Genetics*. St. Louis: Mosby; 1976:523-525.
19. Morris SE, Klein MD. *Pre-feeding Skills*. 2nd ed. United States of America: Therapy Skill Builders; 2000.
20. Roizen NJ, Antshel KM, Fremont W, et al. 22q11.2DS Deletion Syndrome: developmental milestones in infants and toddlers. *J Dev Behav Pediatr*. 2007;28:119-124.