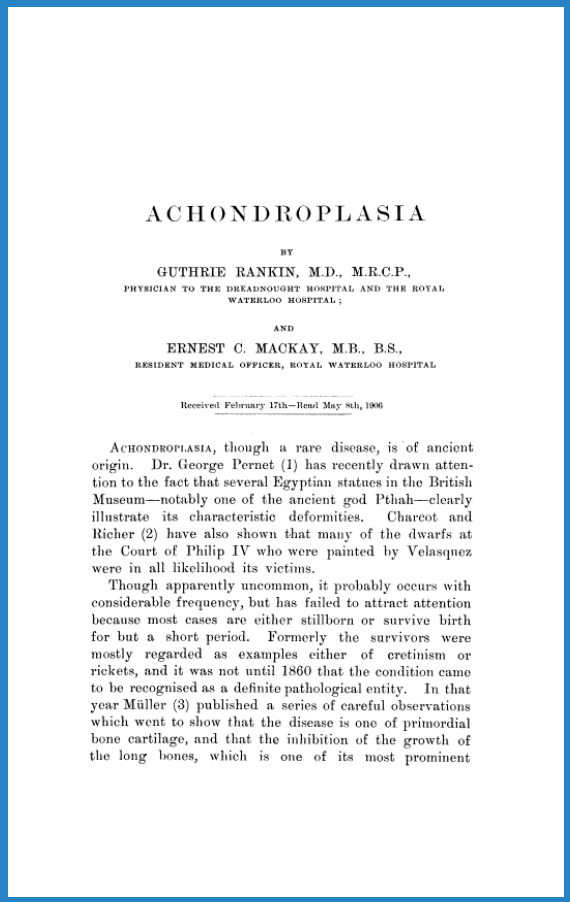
4°Jornadas Nacionales de Auxología y 1°Jornadas Nacionales de Desarrollo
Sociedad Argentina de Pediatría

Desarrollo y estrategias adaptativas en niños con acondroplasia

PABLO CAFIERO

CLÍNICAS INTERDISCIPLINARIAS DEL NEURODESARROLLO
HOSPITAL DE PEDIATRÍA J.P. GARRAHAN



ACHONDROPLASIA

BY

GUTHRIE RANKIN, M.D., M.R.C.P.,
PHYSICIAN TO THE DREADNOUGHT HOSPITAL AND THE ROYAL WATERLOO HOSPITAL;

AND

ERNEST C. MACKAY, M.B., B.S.,
RESIDENT MEDICAL OFFICER, ROYAL WATERLOO HOSPITAL

Received February 17th—Read May 8th, 1906

ACHONDROPLASIA, though a rare disease, is of ancient origin. Dr. George Pernet (1) has recently drawn attention to the fact that several Egyptian statues in the British Museum—notably one of the ancient god Pthah—clearly illustrate its characteristic deformities. Charcot and Richer (2) have also shown that many of the dwarfs at the Court of Philip IV who were painted by Velasquez were in all likelihood its victims.

Though apparently uncommon, it probably occurs with considerable frequency, but has failed to attract attention because most cases are either stillborn or survive birth for but a short period. Formerly the survivors were mostly regarded as examples either of cretinism or rickets, and it was not until 1860 that the condition came to be recognised as a definite pathological entity. In that year Müller (3) published a series of careful observations which went to show that the disease is one of primordial bone cartilage, and that the inhibition of the growth of the long bones, which is one of its most prominent

Med Chir Trans. 1906; 89: 395–418

1. Origen congénito
2. Macrocefalia
3. Depresión del puente nasal
4. Prognatismo
5. Freno del crecimiento de huesos largos con curvatura exagerada
6. Normal desarrollo del tronco
7. Agrandamiento óseo por cambios diafisarios y epifisarios
8. Descentralización del punto medio del cuerpo
9. Manos en tridente
10. Exceso de tejido adiposo
11. Abdomen protuberante
12. Lordosis
13. Piel suave
14. Condición mental normal
15. Tendencia a otras malformaciones congénitas

Evaluación Interdisciplinaria en Acondroplasia
Relación genotipo-fenotipo

- Aspectos clínicos y radiológicos específicos
- Diagnóstico molecular/Asesoramiento por riesgo de recurrencia
- Guías de manejo clínico (AAP-1995/2005)
- Tablas específicas de crecimiento
- **Endofenotipo del desarrollo**

AMERICAN ACADEMY OF PEDIATRICS

CLINICAL REPORT
Guidance for the Clinician in Rendering Pediatric Care

Tracy L. Trotter, MD; Judith G. Hall, OC, MD; and the Committee on Genetics

Health Supervision for Children With Achondroplasia

ABSTRACT. Achondroplasia is the most common condition associated with disproportionate short stature. Substantial information is available concerning the natural history and anticipatory health supervision needs in children with this dwarfing disorder. Most children with achondroplasia have delayed motor milestones, problems with persistent or recurrent middle-ear dysfunction, and bowing of the lower legs. Less often, infants and children may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper-airway obstruction, or thoracolumbar kyphosis. Anticipatory care should be directed at identifying children who are at high risk and intervening to prevent serious sequelae. This report is designed to help the pediatrician care for children with achondroplasia and their families. ***Pediatrics*** **2005;116:771–783;** ***achondroplasia, short stature, children, health supervision.***

ABBREVIATIONS. OFC, occipital-frontal circumference; CT, computed tomography.

INTRODUCTION

This clinical report is designed to assist the pediatrician in caring for children with achondroplasia confirmed by radiographs and physical features. Although pediatricians usually first see children with achondroplasia during infancy, occasionally they are called on to advise a pregnant woman who has been informed of the prenatal diagnosis of achondroplasia or asked to examine a newborn to help establish the diagnosis. Therefore, this report offers advice for these situations as well.

Substantial new information has appeared since publication of the first policy statement on health supervision of children with achondroplasia.[1] In particular, a great deal has been learned about the molecular genetics of the disorder.[2] In addition, a more complete understanding of how certain serious complications can be minimized or avoided has accrued.[3] The new information is incorporated into this report, which is a revision of the original policy statement.

Achondroplasia is the most common condition associated with severe disproportionate short stature.[4] The diagnosis can usually be made on the basis of clinical characteristics and very specific features on radiographs, which include contracted base of the skull, square shape of the pelvis with a small sacrosciatic notch, short pedicles of the vertebrae, rhizomelic (proximal) shortening of the long bones, trident hands, a normal-length trunk, proximal femoral radiolucency, and (by midchildhood) a characteristic chevron shape of the distal femoral epiphysis. Other rhizomelic dwarfing disorders such as hypochondroplasia and thanatophoric dysplasia are part of the differential diagnosis, but achondroplasia usually can be distinguished from them because the changes in hypochondroplasia are milder and the changes in thanatophoric dysplasia are much more severe and invariably lethal. Achondroplasia is an autosomal dominant disorder, but approximately 75% of cases represent new dominant mutations. Achondroplasia is caused by mutation in the gene that codes for the fibroblast growth factor receptor type 3 (*FGFR3*).[5–7] Because virtually all of the causal mutations occur at exactly the same place within the gene,[7] molecular testing is straightforward. It is not necessary to perform molecular testing in every child with a clinical diagnosis of achondroplasia. However, *FGFR3* testing should be performed in children who are in any way atypical or in circumstances in which differentiation from similar disorders, such as hypochondroplasia, is not certain. Such children also should be referred for clinical genetic evaluation.

A great deal is known about the natural history of achondroplasia that can be shared with the family.[3,8] The average adult height in achondroplasia is approximately 4 ft for men and women (Figs 1 and 2).[9] The most common complication, occurring in adulthood, is related to lumbosacral spinal stenosis with compression of the spinal cord or nerve roots.[10,11] This complication is usually treatable by surgical decompression if it is diagnosed at an early stage.

Most children with achondroplasia do well. However, children affected with achondroplasia commonly have delayed motor milestones (Fig 3),[12,13] otitis media, and bowing of the lower legs.[14] Less commonly, infants and children may have serious health consequences related to hydrocephalus, craniocervical junction compression, upper-airway obstruction, or thoracolumbar kyphosis. Although they are less common, anticipatory care should be directed at identifying children who are at high risk and intervening to prevent serious sequelae. Most individuals with achondroplasia are of normal intelligence and are able to lead independent and pro-

The guidance in this report does not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

doi:10.1542/peds.2005-1460
PEDIATRICS (ISSN 0031 4005). Copyright © 2005 by the American Academy of Pediatrics.

PEDIATRICS, Vol. 116 N°3 September 2005

Clinical management of achondroplasia

M J Wright,[1] M D Irving[2]

[1]Northern Genetics Service, Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne, UK
[2]Department of Clinical Genetics, Guy's and St Thomas' NHS Foundation Trust, London, UK

Correspondence to
Dr M J Wright, Northern Genetics Service, Institute of Human Genetics, Newcastle upon Tyne Hospitals NHS Foundation Trust, Central Parkway, Newcastle upon Tyne NE1 3BZ, UK;
michael.wright@nuth.nhs.uk

Accepted 16 March 2011

ABSTRACT
Achondroplasia, one of the skeletal dysplasias and the commonest form of disproportionate short stature, has a different developmental and growth profile compared to average stature children. In addition, a specific group of complications occur more frequently in children with achondroplasia. These include common but usually relatively minor complications such as recurrent otitis media, and rarer but more severe problems such as cervicomedullary compression. Knowledge of these complications, appropriate surveillance strategies and treatment options is essential for the paediatrician. The authors review the published literature in this area and provide a system based approach to the management of the healthcare needs of the child with achondroplasia.

INTRODUCTION

Achondroplasia is the most common form of disproportionate short stature and one of the skeletal dysplasias, a heterogeneous group of several hundred conditions.[1] With a prevalence in newborn infants of 1 in 10 000–30 000, many paediatricians can expect to encounter affected children at some point.[2 3] Although most will be healthy without ongoing health concerns, routine surveillance to detect the significant complications that occur in approximately 10% of these children is required, with appropriate onward referral for specialist intervention.[4] As such, detailed knowledge of the possible complications and a sound approach to anticipatory management can significantly reduce the associated morbidity and even mortality.[5 6] Here we provide an overview of the clinical aspects of achondroplasia and suggest a system based approach to monitor the development of complications.

AETIOLOGY

Achondroplasia is caused in over 95% of cases by one of two recurrent mutations in the gene encoding fibroblast growth factor receptor type 3 (*FGFR3*), both resulting in the same amino acid substitution (G380R).[7 8] These activating mutations have a detrimental effect on longitudinal growth through increased signal transduction in the cartilage growth plate.[9] *FGFR3* is also important in craniofacial, vertebral and neurological development such that this mutation has multiple effects in an affected individual.[10]

Achondroplasia is a dominantly inherited condition with a high new mutation rate, such that 80% of children with achondroplasia have parents of average stature.[11] Of note, there is a strong association between achondroplasia and increasing paternal age. This is thought to be due to a selective growth advantage in sperm containing the *FGFR3* mutations associated with achondroplasia.[12]

DIAGNOSIS

Accurate diagnosis of skeletal dysplasia in children, particularly in newborn babies, while often challenging, is required to anticipate physical complications and provide appropriate genetic counselling. A full skeletal survey (box 1) should be performed if there is clinical suspicion of skeletal dysplasia, such as disproportionate short stature, limb malalignment or specific dysmorphic features. There are key characteristic clinical and radiological signs of achondroplasia that allow it to be distinguished from other conditions (box 1 and figure 1). Confirmatory molecular analysis to detect the recurrent G380R *FGFR3* mutations is available and may be helpful where there is residual doubt about the diagnosis.

GENETIC COUNSELLING

For couples of average stature who have a child with achondroplasia, the risk of recurrence is small (<1%) but not negligible, reflecting the risk of germline mosaicism.[13] The risk is 50% when at least one parent has the condition and there is a 25% risk of homozygous achondroplasia if both parents are affected. This perinatally lethal condition results in severe respiratory compromise secondary to much more severe skeletal dysplasia. Prenatal diagnosis and preimplantation genetic diagnosis are possible, after appropriate genetic counselling, where appropriate facilities exist.

ANTICIPATORY SURVEILLANCE

The American Academy of Pediatrics (http://www.aap.org) has provided specific recommendations for the management of children with achondroplasia.[5] Here we summarise the different areas upon which to focus routine surveillance in line with UK practices.

'NORMAL' GROWTH AND DEVELOPMENT

In assessing the growth and development of a child with achondroplasia, it is important that appropriate comparisons are used. Children with achondroplasia have a different schedule of developmental milestone attainment and their progress should be assessed accordingly.[14 15] Gross motor skills in particular develop later in the child with achondroplasia: approximately 50% of children will sit alone by 9 months and just over 50% will walk alone by 18 months. Similarly, condition-specific centile charts are available to monitor the growth of children with achondroplasia (available online at http://www.lpaonline.org) who have

Arch Dis Child. 2011. doi: 10.1136/adc.2010.189092

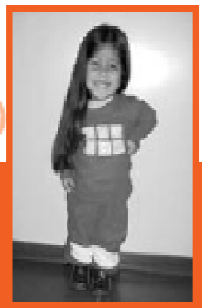
Ficha de Seguimiento de pacientes con Acondroplasia

EDAD	RN	3	6	9	12	18	24	3	1 x año hasta 18	ALTA
RX	*									
Consejo genético	*				*					*
Evaluación antropomética	*	*	*	*	*	*	*		*	*
Evaluación clínica: - Fuerza muscular - R.O.T.	*	*	*	*	*	*	*		*	*
Desarrollo - PRUNAPE	*	*	*	*	*	*	*	*	5 años	
Lenguaje					*		*	*		
Audición							*	*		
Otoscopía			*	*	*	*	*	*		
Investigar: - Ronquido/Apneas - OTR					*		* *	* *		
Ortopedia					*		*	*	*	*
Odontología									5 años	
Neurología - TAC / RMN - PESS										

Adaptado de Health Supervision for Children with Achondroplasia. Comittee on Genetics. Pediatrics Vol. 95 N°3 March 1995

Acondroplasia

Información para padres y pacientes



SERVICIO DE
CRECIMIENTO Y DESARROLLO

HOSPITAL DE PEDIATRIA
"PROF. DR. JUAN P. GARRAHAN"

HOSPITAL GARRAHAN

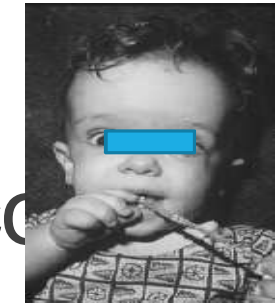
Hospital de Pediatría
Garrahan

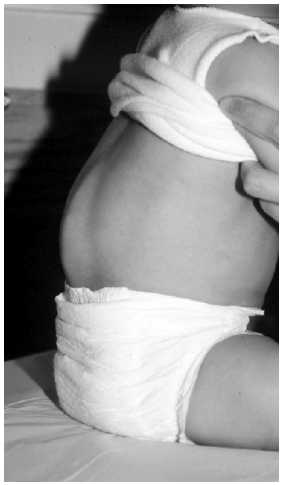
INFORMACIÓN PARA PADRES, FAMILIARES Y PACIENTES

ACONDROPLASIA

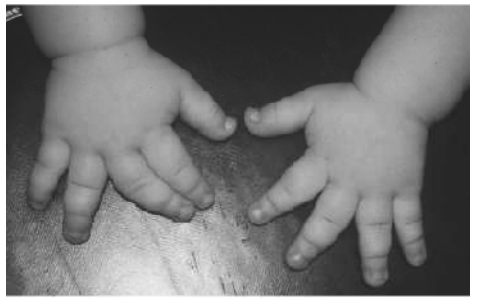
Pichincha 1890 - CABA - C1245AAM - Tel.: (011) 4308-4300 - www.garrahan.gov.ar

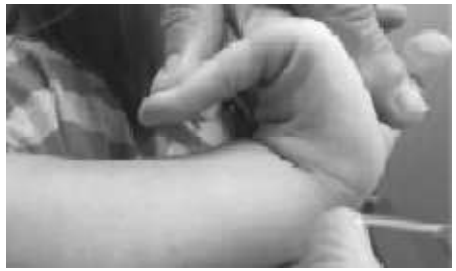
Fenotipo físico





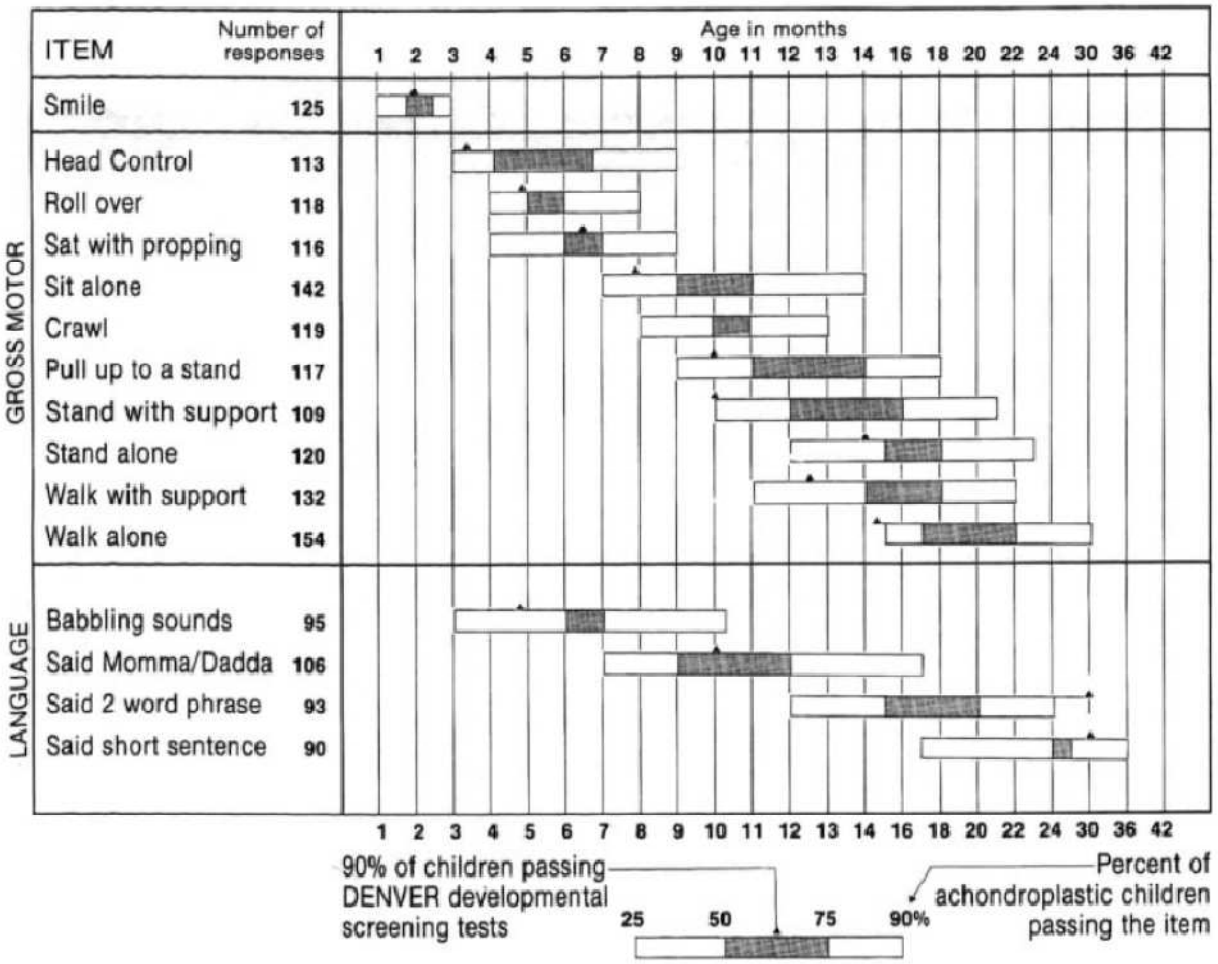
- Baja talla/Acortamiento rizomélico
- Macrocefalia/bossing frontal
- Hipoplasia mediofacial
- Tórax pequeño
- Cifosis toracolumbar/Hiperlordosis lumbar
- Extensión limitada de los codos
- Dedos cortos y manos en tridente
- Hipermovilidad de caderas y rodillas
- Desviación del segmento medial de las piernas
- Hipotonía





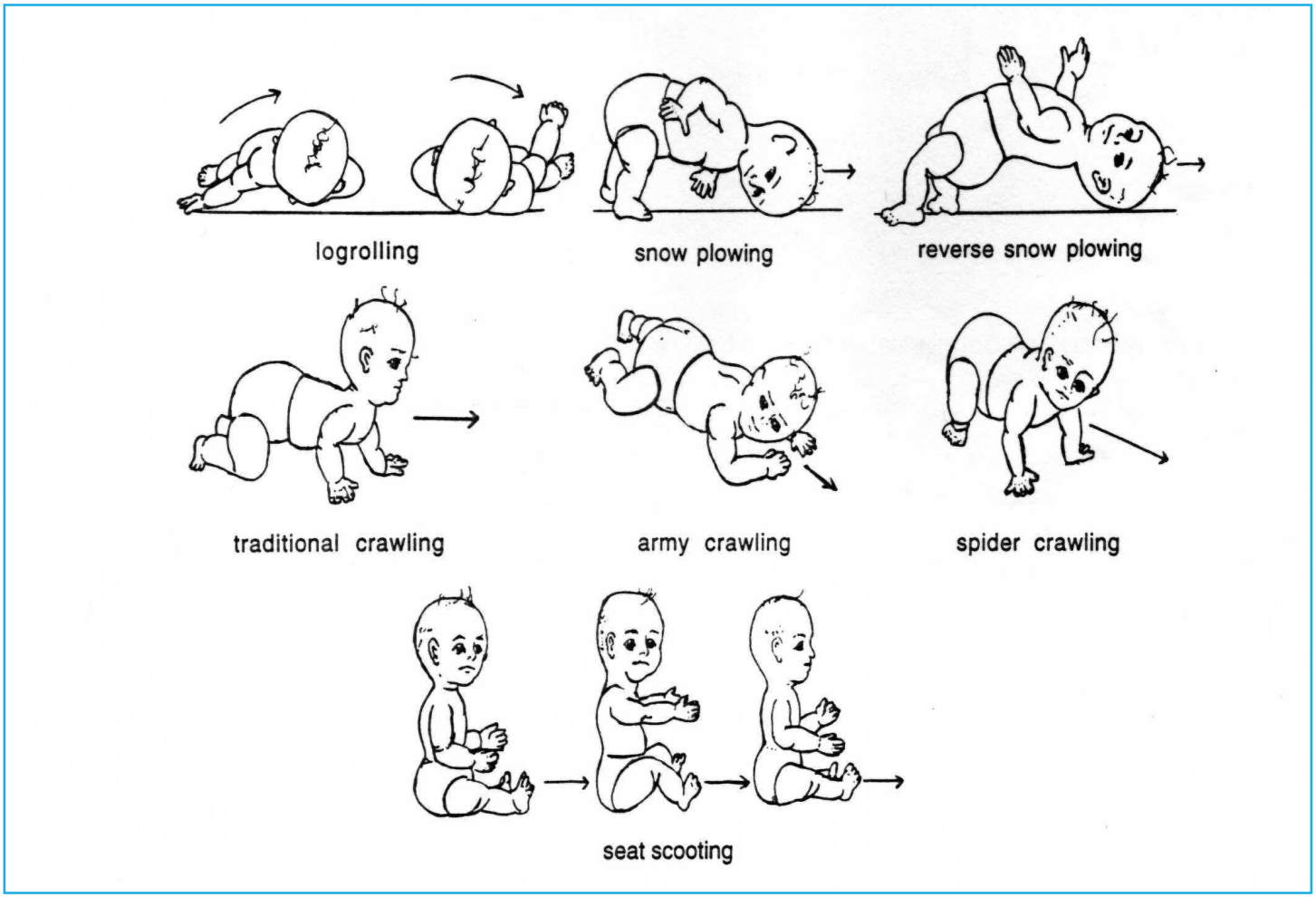
Complicaciones médicas y desarrollo

- Estrechez cervicomedular sintomática del foramen magnum
- Hidrocefalia sintomática
- Disfunción de oído medio/hipoacusia conductiva/hipertrofia adenotonsilar/apneas obstructivas/SAOS
- Compromiso respiratorio restrictivo
- Trastornos ortopédicos
- Maloclusión/superposición dental (hipoplasia maxilar, sobrecrecimiento relativo mandibular)



Todorov. Am J Med Genet 1981; 9: 19

Estrategias motoras adaptativas



Fowler, E. et al. Biophysical Basis for Delayed and Aberrant Motor Development in Young Children with Achondroplasia. Journal of Developmental and Behavioral Pediatrics, Junio, 1997

TABLE 1. Parental Responses to the Movement Strategy Questionnaire (n = 35)

Movement Strategy	Never Seen (n = 35)	%	Age Range (mo)	Average Age Seen (mo)[a]	Ever Predominant Movement (n = 35)	%	Age Range (mo)	Average Age for Predominant Movement (mo)[a]
Logrolling	28	80	2–18	8.4	17	49	2–18	8.4
Traditional crawling	7	20	7–14	9.6	5	14	7–14	10.1
Snow plowing	21	60	6–60	12.2	10	29	8–18	10.1
Reverse snow plowing	15	43	4–72	12.2	3	9	4–14	6.5
Army crawling	27	77	6–78	16.6	23	66	6–30	13.1
Spider crawling	8	23	6–60	17.6	2	6	6–12	9.0
Seat scooting	18	51	6–60	17.7	3	9	9–24	18.3

Fowler, E. et al. Biophysical Basis for Delayed and Aberrant Motor Development in Young Children with Achondroplasia. Journal of Developmental and Behavioral Pediatrics, Junio, 1997

Pautas motoras en pacientes con acondroplasia

Milestone	Sample Size	Percentile (Mo) 25th	50th	75th	90th
Gross motor					
Sit, Head steady	6	10	11	12	13
Roll over	38	4.5	7	10	20
Sit, supported[a]	10	9.5	12	13	14
Sit, no support	37	9	12	16	20.5
Crawl[a]	24	9	12	13.5	14.5
Pull to stand	38	12	15	19	20
Stand, holding on	12	12	14	18	19
Stand, alone	21	16	19	23.5	29
Walk, supported[a]	27	14	17	20	27
Walk, alone	24	14	18	19	22.5
Fine motor					
Reaches	15	6	8	12	15
Pass cube	21	8.5	11	12	14
Take 2 cubes	16	8	10	12	13.5
Thumb-finger grasp	16	11	12	13.5	15
Bang 2 cubes	21	9	11	13	14
Scribbles	23	15	20	27	30
Tower of 2 cubes	16	17.5	22	27	29.5
Tower of 4 cubes	12	25.5	28	31.5	34
Tower of 6 cubes	11	23	24	33	35
Copy circle	13	26.5	39	46.5	48

Fowler, E. et al. Biophysical Basis for Delayed and Aberrant Motor Development in Young Children with Achondroplasia. Journal of Developmental and Behavioral Pediatrics, Junio, 1997

	Acondroplasia		PRUNAPE	
	Mediana	Rango	Percentilo 50	Percentilo 90
Sostén cefálico (26)	6	2-24	1.6	2.63
Sentado sin sostén (23)	9	6-19	7.4	8.1
Camina solo (46)	18	10-52	12.9	15.6

Fano, V., Lejarraga, H. Hallazgos frecuentes en la atención clínica de 96 niños con Acondroplasia.
Arch Argent Pediatr 2000; 98 (6):368-375.

Objetivos de la evaluación del desarrollo en Acondroplasia

Desarrollo motor, comunicación, interacción social y adquisición de habilidades de vida diaria (compromiso espectral)

Observación de la aparición de pautas motoras adaptativas

Detección de las complicaciones médicas asociadas que puedan tener impacto en el desarrollo

Recomendaciones a los padres sobre las características distintivas del desarrollo de estos niños

Valoración del impacto funcional y socio-emocional

Escolaridad

Actividades extracurriculares deportivas/artísticas

Calidad de vida

Necesidades adaptativas (utensilios, útiles escolares, mobiliario, vestimenta, etc)

Transiciones/transferencia

Recomendaciones para la evaluación y seguimiento del desarrollo de niños con Acondroplasia

Los niños con Acondroplasia tienen en su mayoría **inteligencia normal**

Tienen un **desarrollo distintivo y adaptativo específico de la condición**

El examen neurológico es de relevancia para **descartar patología compresiva**

El grado de **hipotonía marca el "tempo"** de adquisición de las pautas motoras gruesas.

El nivel cognitivo no guarda relación con el retraso motor, pero **sí con el compromiso respiratorio** severo **o** las **complicaciones neurológicas**

El desarrollo del **lenguaje debe ser evaluado periódicamente** por el pediatra

La **actividad física escolar no debe ser restringida**, salvo deportes de alto impacto

Considerar el **impacto funcional y socio-emocional** en todas las edades

2011

DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY ORIGINAL ARTICLE

Functional performance in young Australian children with achondroplasia

PENELOPE JANE IRELAND[1] | JAMES MCGILL[2] | ANDREAS ZANKL[3] | ROBERT S WARE[4] | VERITY PACEY[5] | JENNY AULT[6] | RAVI SAVARIRAYAN[7] | DAVID SILLENCE[8] | ELIZABETH M THOMPSON[9] | SHARRON TOWNSHEND[10] | LEANNE MARIE JOHNSTON[1]

1 School of Health and Rehabilitation Sciences, The University of Queensland, Brisbane, Queensland. **2** Department of Metabolic Medicine, Royal Children's Hospital, Brisbane, Queensland. **3** UQ Centre for Clinical Research, The University of Queensland, Brisbane, Queensland. **4** School of Population Health, The University of Queensland, Brisbane, Queensland. **5** Department of Physiotherapy, The Children's Hospital at Westmead, Sydney, New South Wales. **6** Department of Paediatric Rehabilitation, The Children's Hospital at Westmead, Sydney, New South Wales. **7** Victorian Clinical Genetics Service, Royal Children's Hospital, Melbourne, Victoria. **8** Department of Clinical Genetics, The Children's Hospital at Westmead, Sydney, New South Wales. **9** SA Pathology, South Australian Clinical Genetics Service, Women's and Children's Hospital, Adelaide, South Australia. **10** Genetics Service of Western Australia, King Edward Memorial Hospital, Perth, Western Australia, Australia.

Correspondence to Penelope J Ireland, Senior Physiotherapist at Queensland Paediatric Rehabilitation Service, Royal Children's Hospital, Herston Road, Herston 4029, Brisbane, Queensland, Australia.
E-mail: penny_ireland@health.qld.gov.au

2012

DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY ORIGINAL ARTICLE

Development in children with achondroplasia: a prospective clinical cohort study

PENELOPE J IRELAND[1] | SAMANTHA DONAGHEY[1] | JAMES McGILL[2] | ANDREAS ZANKL[3] | ROBERT S WARE[4,5] | VERITY PACEY[6] | JENNY AULT[7] | RAVI SAVARIRAYAN[8] | DAVID SILLENCE[9] | ELIZABETH THOMPSON[10] | SHARRON TOWNSHEND[11] | LEANNE M JOHNSTON[12]

1 Queensland Paediatric Rehabilitation Service, Royal Children's Hospital, Brisbane, Qld. **2** Department of Metabolic Medicine, Royal Children's Hospital, Brisbane, Qld. **3** University of Queensland Centre for Clinical Research, Royal Brisbane and Women's Hospital, Brisbane, Qld. **4** School of Population Health, The University of Queensland, Brisbane, Qld. **5** Queensland Children's Medical Research Institute, Brisbane, Qld. **6** Department of Physiotherapy, The Children's Hospital at Westmead, Sydney, NSW. **7** Department of Paediatric Rehabilitation, The Children's Hospital at Westmead, Sydney, NSW. **8** Victorian Clinical Genetics Service, Royal Children's Hospital, Melbourne, Vic. **9** Department of Clinical Genetics, The Children's Hospital at Westmead, Sydney, NSW. **10** SA Pathology, South Australia Clinical Genetics Service, Women's and Children's Hospital, Adelaide, SA. **11** Genetics Service of Western Australia, King Edward Memorial Hospital, Perth, WA. **12** School of Health and Rehabilitation Sciences, The University of Queensland, Brisbane, Qld, Australia.

Correspondence to Mrs Penelope J Ireland at Queensland Paediatric Rehabilitation Service, Royal Children's Hospital, Herston Road, Herston 4029, Brisbane, Qld, Australia.
E-mail: penny_ireland@health.qld.gov.au

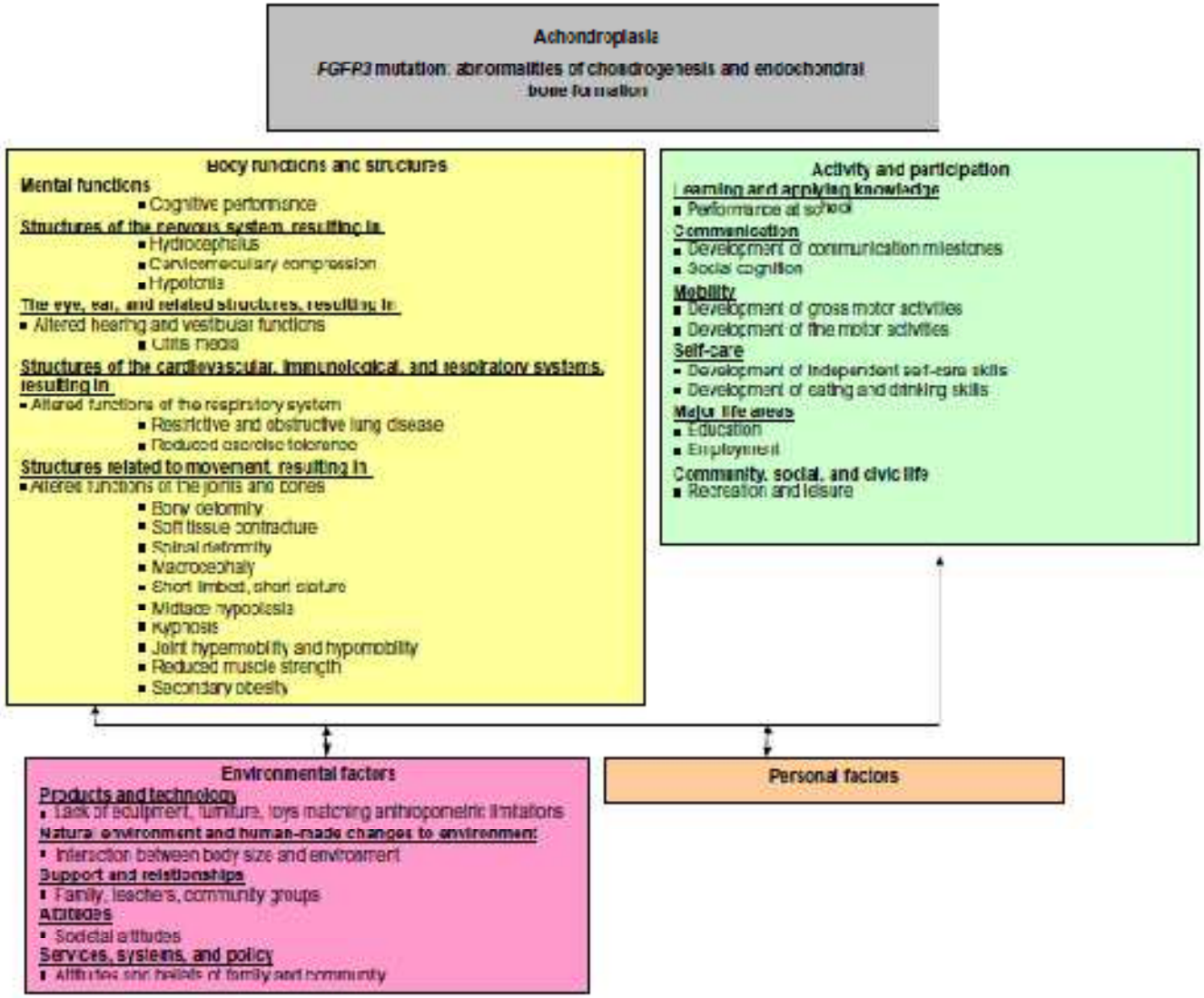


Figure 1 ICF model of achondroplasia.

Abbreviations: *FGFR3*, fibroblast growth factor receptor 3; ICF, the International Classification of Functioning, Disability, and Health.

Ireland P et al. Optimal management of complications associated with achondroplasia, The Application of Clinical Genetics, 2014.

Table I: The 90th centile age of achievement (mo) for gross motor, fine motor, communication, and feeding skills for children with achondroplasia

Milestone	*n*	Median (min–max)	90th centile this study (*n*=48 all items)	90th centile Fowler et al. 1997 (*n* for item)	90th centile Denver II test
Gross motor					
Lift head when lying on stomach[a]	45	4.5 (0.5–15.5)	7		3.5
Roll over[a]	45	5.3 (1–15.5)	9.9	20 (38)	5.2
Snow plough	11	12 (8–36)	21		
Reverse snow plough	22	8 (3–16)	13.8		
Commando crawl	45	9 (5.75–18)	14.2		
Bear walking	21	12 (6–21)	18		
Traditional crawling[a]	16	12 (6–24)	18.3	14.5 (24)	
Into sitting from lying[a]	43	14.3 (9–32)	18.5		10
Into sitting from standing	39	15 (8–32)	22.8		
Into standing from sitting[a]	41	15 (8.5–32)	20	20 (38)	10
Stand holding on[a]	44	13.5 (6–32)	20	19 (12)	12
Stand unsupported	39	17 (9–35.5)	24	29 (21)	14
Walk holding on[a]	45	16 (8.8–32)	22	27 (27)	12.7
Walk independently[a]	44	19.4 (13.5–46)	26	22.5 (24)	15
Fine motor					
Reach for object[a]	46	4 (1–10)	6.5	15 (15)	5.5
Pass objects[a]	45	5.5 (3–15)	10	14 (21)	7.5
Bang objects together	42	8 (4–15)	11.1	14 (21)	11
Scribble with crayon[a]	42	14.5 (5.5–26)	20	30 (23)	17
Unscrew lid from jar	36	22.5 (12.5–42)	35		
Draw circle[a]	36	27 (17–36)	36	48 (13)	45
Build tower two blocks[a]	43	16 (7–54)	20.9	29.5 (16)	20
Build tower eight blocks[a]	38	22.5 (12–54)	32.9	35 (11)	42
Communication					
Smile[a]	46	1.5 (0.3–3.5)	2.8		2
Babble[a]	46	7.6 (2–16.5)	11.3		9
Wave[a]	45	10 (6–20)	14.3		14
Say 'mama'[a]	47	10 (6–18)	14		13
Shake head	45	13 (5–22)	20		
Peek-a-boo	45	10 (3–20)	13		9.7
1 step request[a]	44	18 (10–30)	23.5		
Identify body parts	46	16.7 (9–42.5)	21		27
Imitate words	46	14 (9–26)	22		
Use single words[a]	44	18 (7–36)	22		15
Combine two words[a]	39	24 (11–41)	30		25
Short sentences[a]	37	27 (16–54)	38.4		
Feeding					
Cup drinking[a]	43	10 (4–19)	15.6		17
Puree/smooth solids	48	5.4 (2–9)	6.5		
Mashed solids	47	7 (4–14)	10.2		
Finger feeding	48	9.8 (6–18)	12.3		7
Self-feed with spoon[a]	46	16.5 (7.5–30)	22		20

[a]Denotes those items found in the Australian State Government Personal Health Record Books.

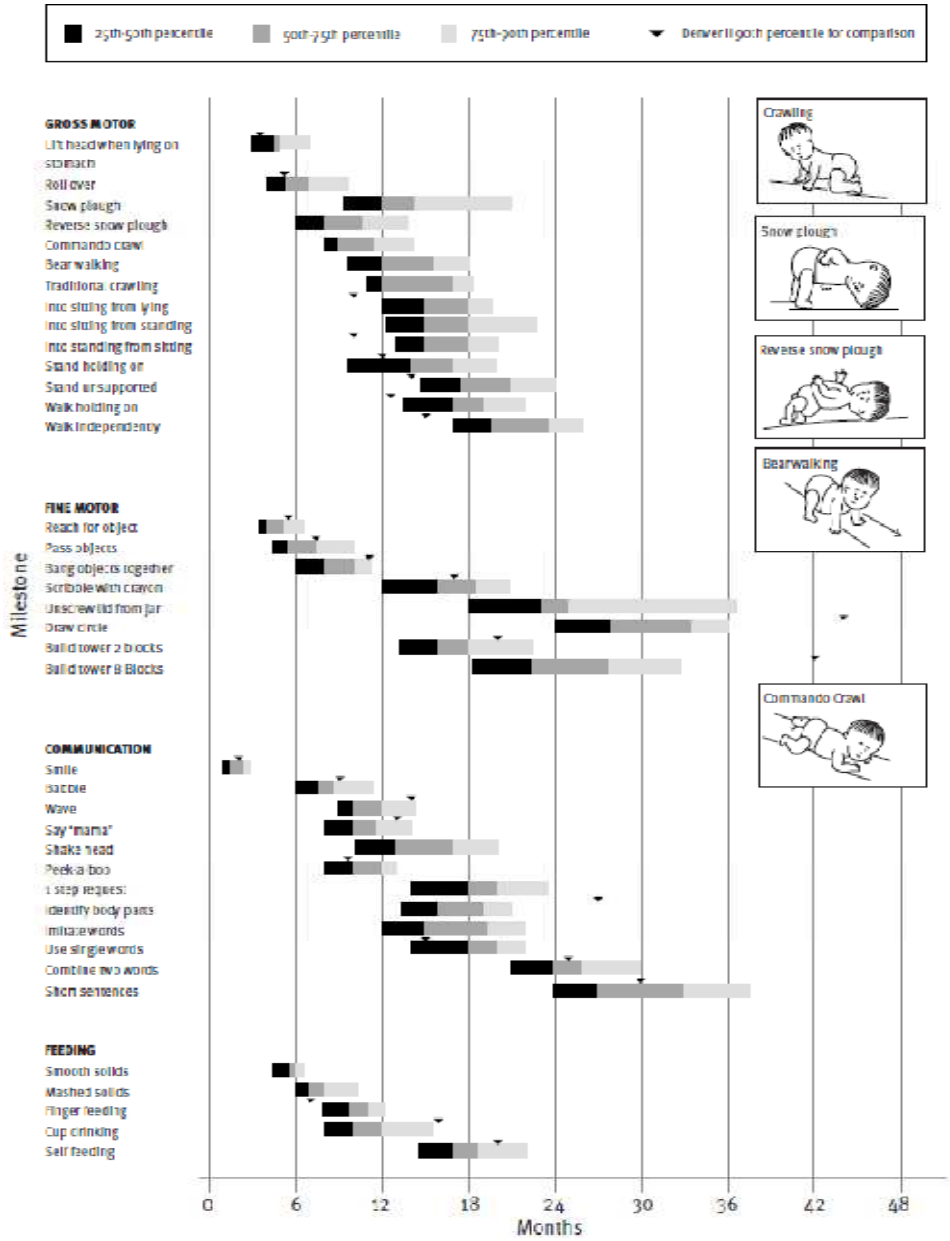
Table II: Preorthograde movement strategies used by children with achondroplasia

	Ireland et al. 2011 (*n*=48)		Fowler et al.[2] (*n*=35)	
Movement strategy[a]	*n*	Mean age (mo)	*n*	Mean age (mo)
Snow plough	11	12	21	12.2
Reverse snow plough	24	8	15	12.2
Commando crawl	48	11	27	16.6
Bear walking	21	12	8	17.6
Traditional crawling	16	13.8	7	9.6

[a]Parents may have reported that children used more than one strategy.

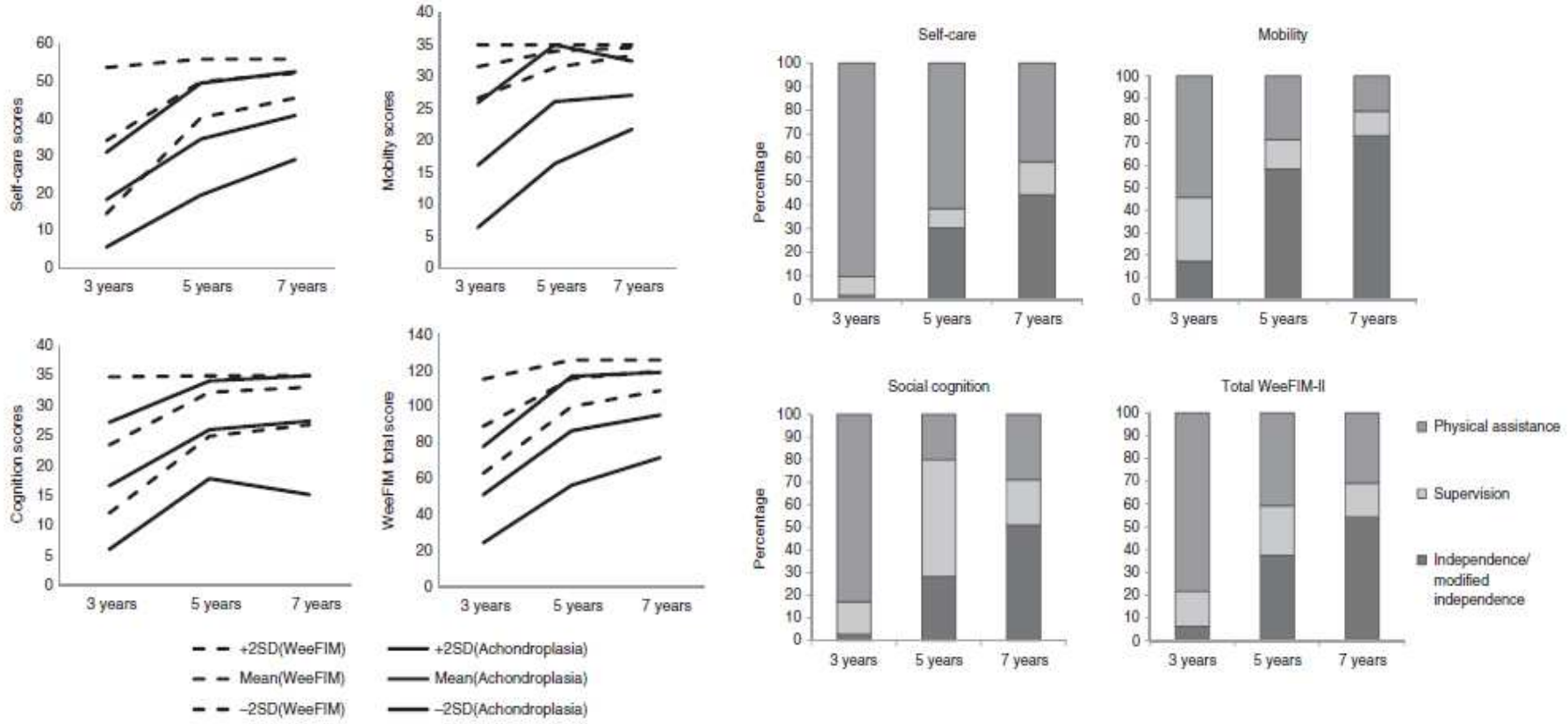
Table III: Transitional movement strategies used by children with achondroplasia

Transition	Movement strategy[a]	Number of children (*n*=48)
Into sitting from lying	Roll onto one side	9
	Push up from stomach	29
	Other	3
From standing up into sitting	Dropping backwards	13
	Squatting down	21
	Spreading legs	8
	Other	3
Pull into standing from sitting down	Kneeling up on both knees	21
	Lying on stomach	19
	Other	6

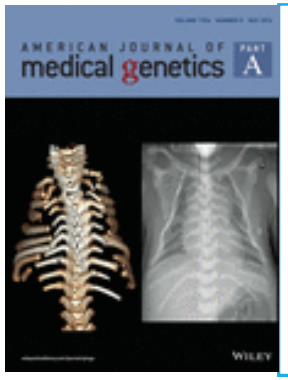


Ireland. Dongahey, McGill, Zanki, Ware, Johnson, Pacey, Ault, Savarirayan, Sillence, Thompson, Townshend, Johnston
Dev Med Child Neurol 2012; 54: 532-537

Evaluación funcional



Evaluación de función neuropsicológica



ORIGINAL ARTICLE

AMERICAN JOURNAL OF
medical genetics

The Neuropsychological Function of Children With Achondroplasia

Kimberley Wigg,[1] Louise Tofts,[2] Suzanne Benson,[1,2] and Melanie Porter[2,3]*

Am J Med Genetics Part A 2016; 170(5): 1-7

Minerva Pediatrica 2016 Feb 19

Cognitive phenotype and language skills in children with achondroplasia

Cinzia GALASSO, Martina SIRACUSANO, Nadia EL MALHANY, Caterina CERMINARA, Mariabernarda PITZIANTI, Monica TERRIBILI ✉
Department of Neuroscience, Paediatric Neurology Unit, "Tor Vergata" University of Rome, Rome, Italy

Minerva Pediatr 2016 Feb 19.

Función neuropsicológica en Acondroplasia

1. Función intelectual
2. Función académica
3. Atención
4. Memoria
5. Función viso-espacial
6. Función ejecutiva
7. Función adaptativa

- CI dentro de límites normales pero con media de CI total: 92 (mayor compromiso a nivel de CI verbal)

Déficits encontrados en las siguientes áreas:

- Función ejecutiva (atención y memoria de trabajo): organización y planificación/resolución de problemas
- Atención visual selectiva y atención auditiva sostenida
- Cambio del foco atencional/Atención dividida
- Respuestas inhibitorias
- Habilidades lectoras y de aritmética
- Trastorno del habla y del lenguaje
- Tendencia a dificultades socio-emocionales (ej: ansiedad, dificultades conductuales)

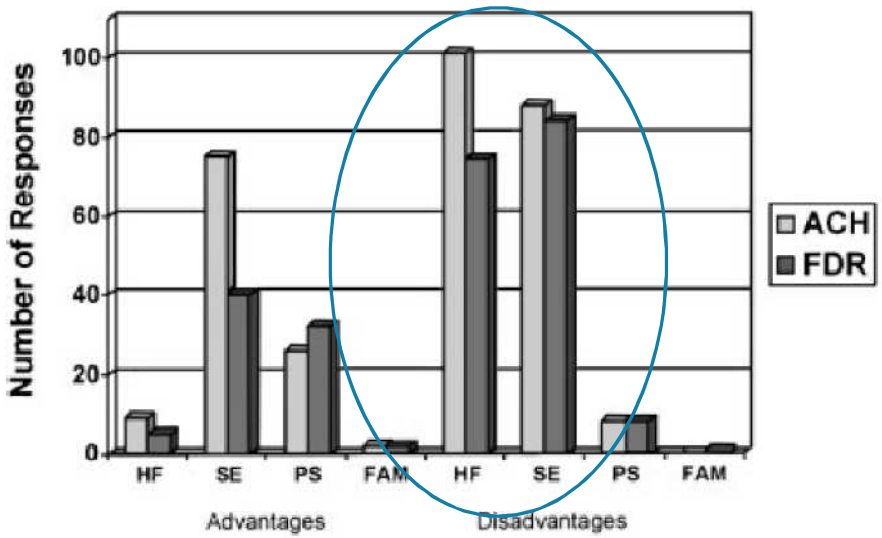
Estrategias preventivas y anticipatorias

- Tener en cuenta la manifestación del problema ante ambientes menos estructurados (**transiciones escolares**)
- Considerar el mayor riesgo para bajo rendimiento académico/trastorno del aprendizaje (**retraso del lenguaje** con déficits en expresión y comprensión verbal, más **dificultades en habilidades lectoras y aritmética**)
- Tener en cuenta la puesta en marcha de **estrategias que maximicen el potencial de aprendizaje**, considerando vulnerabilidades en áreas de atención, función ejecutiva, memoria de trabajo e inteligencia verbal
- Minimizar el **impacto socio emocional**

Calidad de vida en personas con Acondroplasia

Living With Achondroplasia in an Average-Sized World: An Assessment of Quality of Life

Sarah E. Gollust,[1] Richard E. Thompson,[2] Holly C. Gooding,[1] and Barbara B. Biesecker[1]*
[1]Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland
[2]Johns Hopkins University Biostatistics Center, Baltimore, Maryland

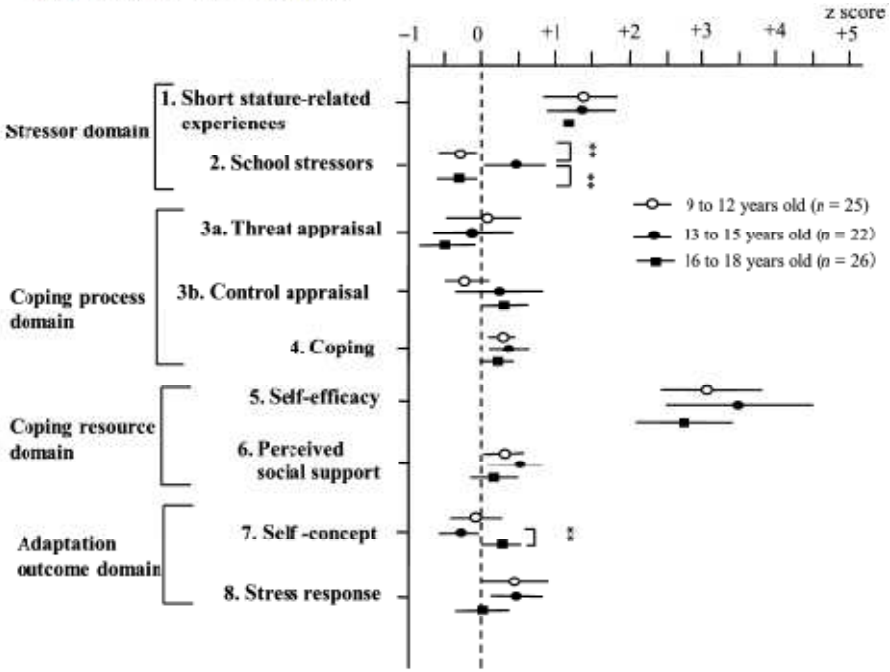


Am J Med Gen 120A; 447-458

ORIGINAL ARTICLE
Journal of Clinical Nursing

Psychosocial profiles of children with achondroplasia in terms of their short stature-related stress: a nationwide survey in Japan

Naoko Nishimura and Keiichi Hanaki



J Clin Nursing 2013; 23: 3045-3056

Acondroplasia:
Desafíos en la agenda a futuro

“No afirmo saber cuánto vale la vida de una persona: no se puede responder con palabras a esa cuestión, sino sólo con obras, con la creación de una sociedad más humana.
Lo único que se es que la sociedad actual desaprovecha, y al hacer prevalecer la hipocresía, vacía la mayoría de las vidas que no destruye; y también que, en los términos de esta sociedad, un médico que no se limita a vender curas, ya sea directamente a sus pacientes o a través de los servicios estatales, es inestimable.”

John Berger- Un hombre afortunado, 1967

MUCHAS GRACIAS