PCHR INSERT FOR BABIES BORN WITH ACHONDROPLASIA

First Edition

These are extra pages for your child's Personal Health Record. They have been created especially for babies and young children with Achondroplasia. They contain background information for families and health care professionals, and guidelines and checklists for growth and development.

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Children with Achondroplasia

guidance for parents and health care professionals

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North West Regional Genetics Service: 0161 276 6506 www.mangen.co.uk

Central Manchester University Hospitals NHS
NHS Foundation Trust

Contents

Different pages in this PCHR insert for Achondroplasia should be filled in by different groups of people. The contents table and key below explain who needs to read or complete each page.

Specialist Doctor Details

Children with Achondroplasia are often looked after by more than one specialist, and this page is a record of all the doctors who are involved in caring for your child. If you visit a specialist doctor who isn't listed, they should add their details below.

Name	Title/Specialty	Address/Phone Number
		☎
		☎
		☎
		☎

Information for You and Your Family

What is Achondroplasia?
Achondroplasia is a genetic condition. It is caused by a fault in a particular gene called FGFR3. Genes are responsible for individual characteristics such as eye and hair colour.

About 1 in 25,000 babies born in the UK will have achondroplasia. In 75% there is no family history of achondroplasia, in the remainder, one or both parents have achondroplasia. A person with achondroplasia has a 1 in 2 , or 50% chance of having an affected child.

How is it Diagnosed?
Usually the diagnosis is straightforward and is made by carefully examining x-rays of the skeleton. If there is any uncertainty about the diagnosis, your doctor may carry out a genetic test to confirm the diagnosis.

How will My Child be Affected?
The main difference found in achondroplasia is short stature, which is due to shortening of the long bones of the legs (femur and tibia). The average adult height for an adult with achondroplasia is between 3'8" (110 cm) and 4'9" (143cm).

A number of rare complications can occur in early life and regular monitoring of your child is important to ensure that problems are identified and appropriately treated. In the section 'Caring for Your Baby' these risks are described in more detail along with everyday handling strategies to minimise these risks.

Though it will take longer for your child to reach developmental milestones such as sitting unsupported, and walking alone, it is important to remember that people with achondroplasia have the same range of intelligence as the general population, and live full and productive lives.

Professional Care
Usually a community or general paediatrician, will have overall responsibility for the care of your child's health. In addition, there may be another key worker who will have a co-ordinating and support role where a number of difference professionals are involved. Your child's key worker is:

Name:

Contact Details:

Caring for Young Babies and Children with Achondroplasia

Important Information for Parents and Carers
A number of rare complications involving the head, neck and spine can occur in early life. Monitoring for these allows early diagnosis and treatment, and may prevent some of the adult complications. Babies and young children with achondroplasia should be handled differently to try and minimise these risks.

Limiting Uncontrolled Head Movements
The head of infants with achondroplasia is relatively large and requires good support until independent sitting is achieved. The foramen magnum (opening through which the spinal cord passes into the base of the skull) is narrower in babies and children with achondroplasia. Uncontrolled head movements can 'squash' the spinal cord causing damage to the nerves and arteries in the spinal canal. A scan will be carried out in early life to look at the size of this opening, and the size of the fluid filled spaces (ventricles) in the brain.

Hydrocephalus
The fluid filled spaces in the brain (ventricles) are large in people with achondroplasia. Your doctor will monitor this by regular measurements and a base-line scan. Sometimes fluid flow can be obstructed and surgery is required. Symptoms which might suggest this and are a reason for urgent referral are: a bulging soft spot, persistent irritability, vomiting or lethargy or apparent headaches.

Avoiding Spinal Curvature
A small gibbus (C' shaped or curved spine) is present in most infants with achondroplasia and will resolve with standing. This becomes fixed in a small number and then contributes to spinal problems in adult life. Research has demonstrated that avoiding unsupported sitting in babies and young children with achondroplasia minimises this risk.

The baby's lower spine is curved. This is more noticeable when the baby is sitting in an upright position.

When the baby lies down the curve flattens out. The is a much better position for babies with achondroplasia.

Providing Adequate Head and Spine Support
The infant with achondroplasia should be nursed on a firm surface as much as possible. Babies must be properly supported when sitting is essential. Soft canvas baby rockers and bouncinettes , baby walkers, carriers and strollers do not provide enough neck and spine support and should not be used.

Car Seats
Should have a firm back and neck support and be rear facing for as long as possible. Seats which adjust to allow babies to lie flat are a good idea for babies.

Play
Playground swings and infant bouncers, the sort you have on a hook from the door frame should be avoided, as should trampolines. Care should be taken with any activity that places a strain on the neck, e.g. baby gymnastics.

Growth
Special growth curves are included in this booklet and should be used to monitor growth. Monitoring weight is also important. Failure to gain adequate weight in infancy can be a sign of an underlying complication and should be investigated. In older children it is important to maintain an appropriate weight as obesity may be a problem in adult life. Healthy eating habits and appropriate regular exercise should be encouraged from an early age. Growth hormone does not appear to make a difference to final adult height.

Dental
Differences in the shape of the face and palate may lead to overcrowding of the teeth and bite abnormalities. An orthodontic assessment should be carried out around the age of five years and repeated subsequently as recommended by the orthodontist.

Orthopaedic Issues
Instability around the knee, bowing backwards of the knee and sideways bowing of the lower limbs is common. This is most obvious in the second year of life. This rarely requires treatment, and usually improves with age. However in the presence of persistent pain on exercise, orthopaedic assessment is needed. Flexion contracture at the hips can occur in infancy and can be minimized with appropriate exercises. The other joints tend to be loose with the exception of the elbows where a mild inability to straighten the arm is common.

Anaesthesia
Throughout life, anaesthesia in people with achondroplasia requires extra care, particularly with positioning of the head and neck and use of appropriate dosages for size. Spinal and epidural anaesthesia should generally be avoided.

Breathing Complications
Snoring is common in young children with achondroplasia and by itself is not something to worry about. Because respiratory problems are common, a screening sleep study will be organised. This should be repeated if you notice any change in breathing patterns such as big pauses between breaths, loud snoring, daytime sleepiness, deep sighs during sleep.

Hearing
The shape of the inner ear is different in achondroplasia and accumulation of fluid in the middle ear (‘glue ear') and ear infections are common. Regular monitoring of hearing is important throughout infancy and childhood. In the presence of language delay, hearing loss should be excluded. Ear examination is recommended in the presence of symptoms suggesting infection.

Psychosocial
Parents may find it helpful to meet other families. Your regional genetics service may facilitate this. The RGA is a good source of support and information. Contact details are on page 20.

Information for Healthcare Professionals

Diagnosis and Follow up
Achondroplasia is the commonest skeletal dysplasia associated with severe disproportionate short stature. It is an autosomal dominant condition occurring in approximately 1 in 25,000 and is due to a mutation in the FGFR3 gene, with most being new dominant mutations. An affected parent has a 50% risk of passing on the gene to offspring. If both parents are affected, there is also a 25% risk of a child inheriting two copies of the faulty gene. This results in a different and more severe skeletal dysplasia.

Diagnosis
The diagnosis is based on characteristic clinical and radiological findings. The X-rays should be reviewed by a paediatric radiologist. Genetic testing is indicated where the diagnosis is unclear or with an atypical phenotype.

The Role of the Regional Genetic Service
A Clinical Geneticist is often involved in the diagnosis of Achondroplasia. They also have a role to play in ensuring children receive appropriate follow up including genetic counselling and education for families and professionals.

Monitoring and Review
Adults and children with achondroplasia have normal intelligence and life expectancy and lead independent and productive lives.

There are some particular health risks that require monitoring and vigilance. The commonest overall is spinal cord stenosis with compression of the spinal cord or nerve roots in adult life. Development of a fixed gibbus in childhood is thought to contribute to this, hence the importance of avoiding unsupported sitting in childhood.

In childhood, the commonest complications are delayed motor milestones and otitis media (glue ear).

Serious complications in early childhood include cranio-cervical junction compression and upper airway obstruction. Investigation and monitoring in early childhood are vital in early diagnosis and treatment of these serious complications. Special care needs to be taken if anaesthesia is necessary for surgery or imaging.

Copies of detailed review guidelines are available from: www.mangen.co.uk

Normal development milestones are included on page 15. Failure to achieve these and/or persistent severe hypotonia or other abnormal neurological signs, failure to thrive, or abnormal breathing patterns warrant URGENT REFERRAL.

Monitoring Your Child's Growth (Birth to 6 months)

Your child's doctor will measure height, weight and head circumference at each visit and compare these with the special growth charts at the back of the booklet.

	Birth		2 Months		4 Months		6 Months	
	Date:		Date:		Date:		Date:	
	cm	centile	cm	centile	cm	centile	cm	centile
Height								
Weight								
Head Circ								
Signature								
Comments								

Monitoring Your Child's Growth (9 to 18 months)

Your child's doctor will measure height, weight and head circumference at each visit and compare these with the special growth charts at the back of the booklet.

	9 Months		12 Months		15 Months		18 Months	
	Date:		Date:		Date:		Date:	
	cm	centile	cm	centile	cm	centile	cm	centile
Height								
Weight								
Head Circ								
Signature								
Comments								

Monitoring Your Child's Growth (2 to 5 years)

Your child's doctor will measure height, weight and head circumference at each visit and compare these with the special growth charts at the back of the booklet.

	2 Years		3 Years		4 Years		5 Years	
	Date:		Date:		Date:		Date:	
	cm	centile	cm	centile	cm	centile	cm	centile
Height								
Weight								
Head Circ								
Signature								
Comments								

Monitoring Your Child's Growth (6 to 9 years)

Your child's doctor will measure height, weight and head circumference at each visit and compare these with the special growth charts at the back of the booklet.

	6 Years		7 Years		8 Years		9 Years	
	Date:		Date:		Date:		Date:	
	cm	centile	cm	centile	cm	centile	cm	centile
Height								
Weight								
Head Circ								
Signature								
Comments								

Key Investigations Checklist

These important checks should all take place within a few weeks of birth. This form should be used to record when these checks took place along with a brief note about the results.

Investigation	**Date**	**Signature**	**Results**	**Comments**
Skeletal Survey				
Pulse Oximetry				
Brain Imaging				
Other				
Hearing Tests				
Newborn				
12 months				
24 months				
4 years				
Other				

Details of Hospital Visits and Other Appointments

Sheet Number

Children with Achondroplasia may be under the care of several specialist doctors and health care professionals. This page provides space for them to record brief details about the appointment and will be a record of who you saw and why.

When (Date)	Where (Hospital/GP etc)	With Who (Name)	Doctors Comments

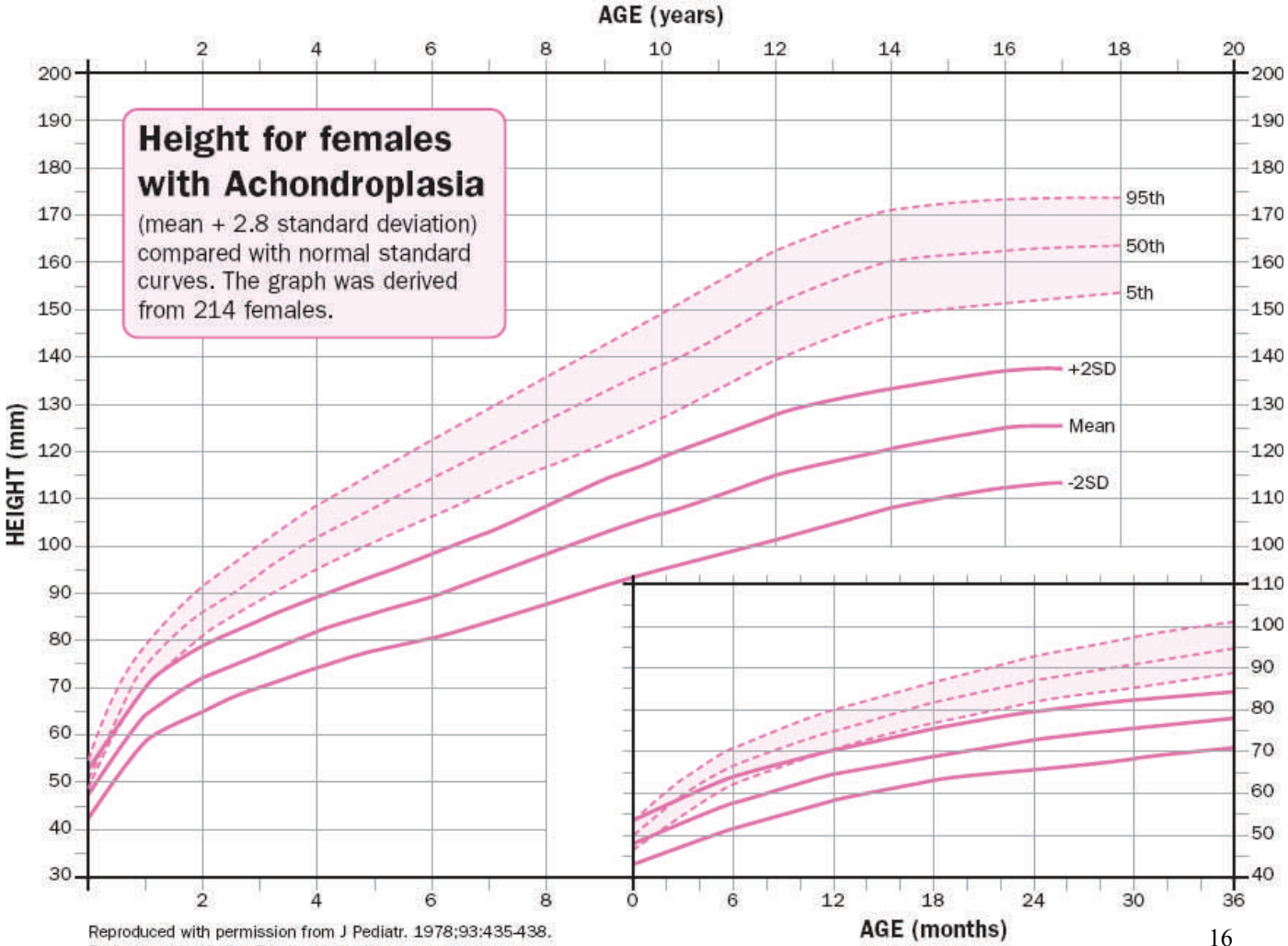
Development Milestones

The vast majority of children with achondroplasia have normal intelligence, and their life goals and aspirations should match those of other family members . It is normal in achondroplasia to take longer to reach developmental milestones. Motor delay is particularly common as illustrated in this chart.

SKILL	25th-90th %ile for children with achondroplasia	Average age for average stature
Sit without a support	9-20.5	5.5
Pull to stand	12-20	7.5
Stand Alone	16-29	11.5
Walk	14-27	12
Reach	6-15	3.5
Pass Object	8-14	6
Bang two objects	9-14	8.5
Scribble	15-30	13.5

Language Development

Speech and language development may be delayed when compared to children without achondroplasia. However, 90% will be babbling by 9 months, saying Mamma or Dadda by 18 months, 2 word phrases by 30 months and speaking in short sentences by three years.



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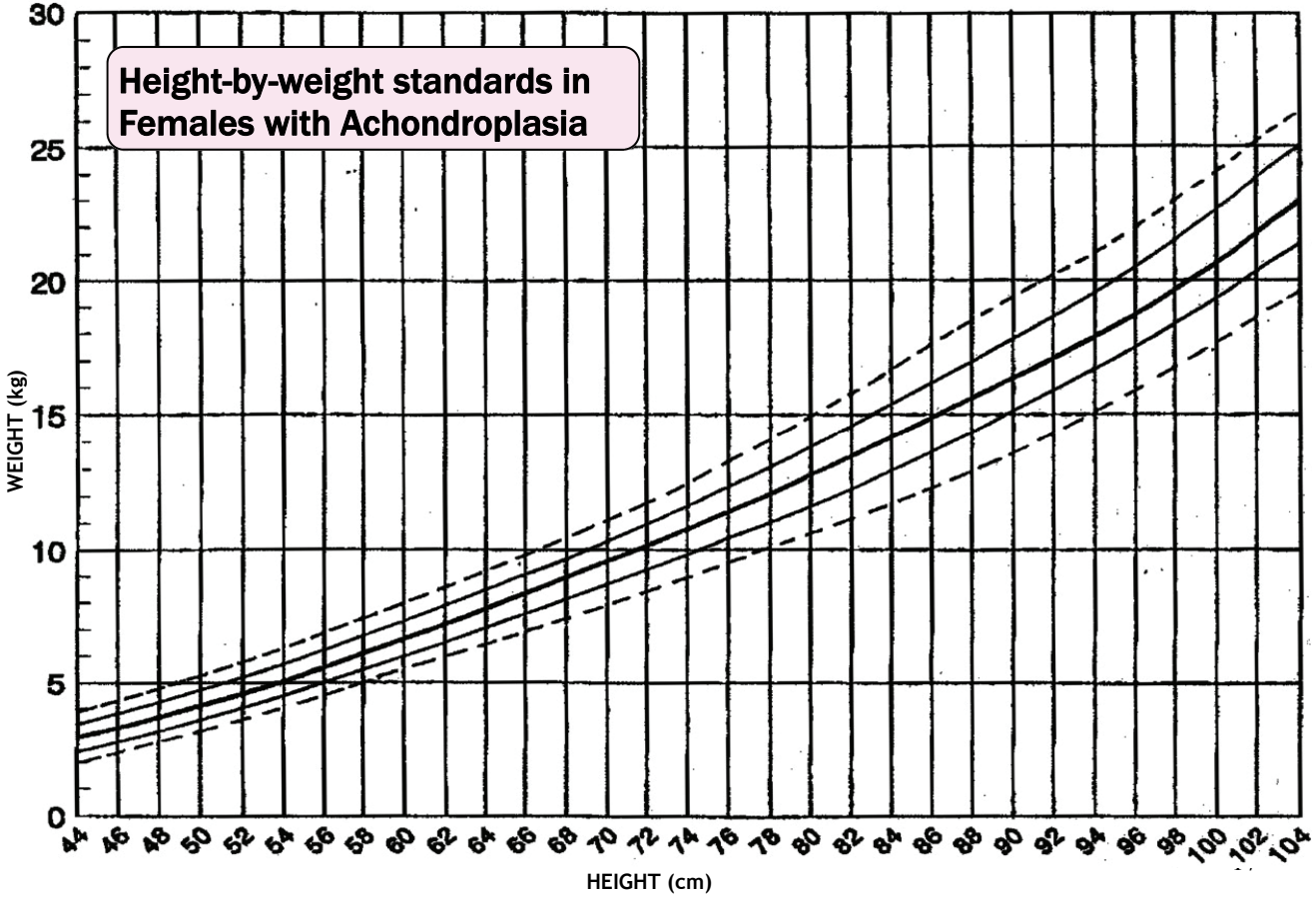
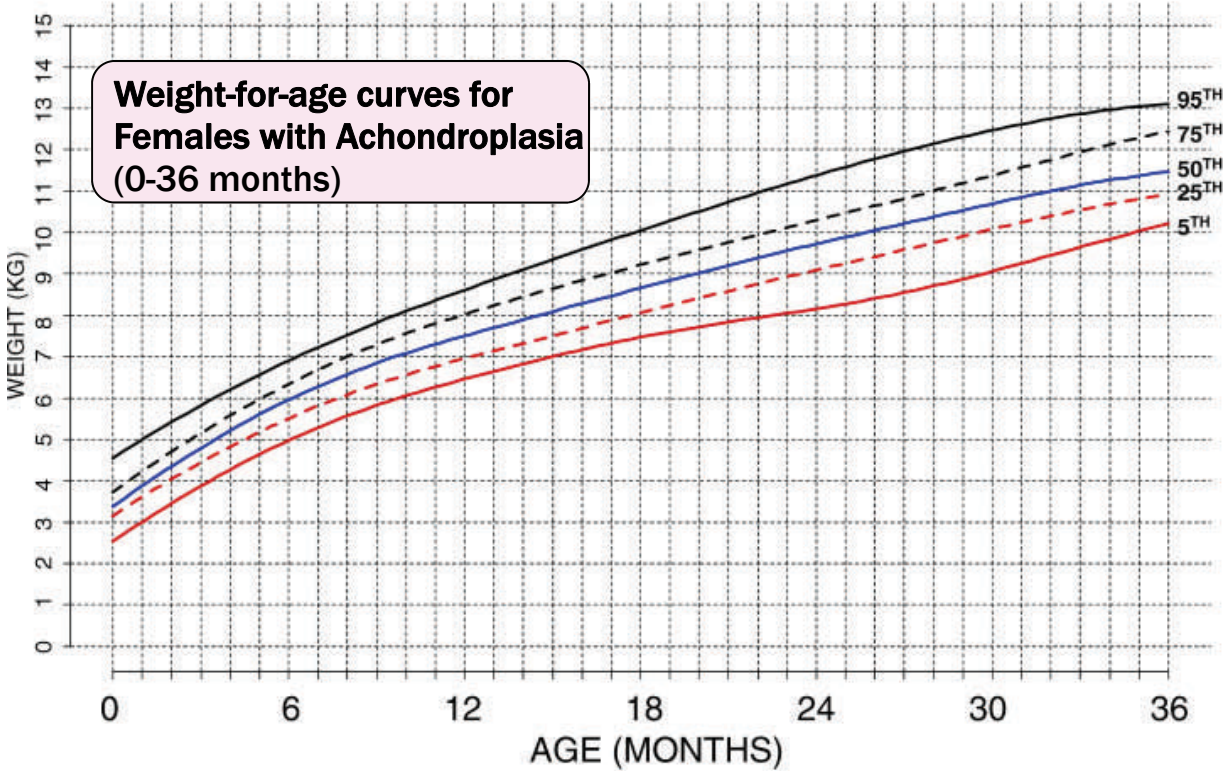
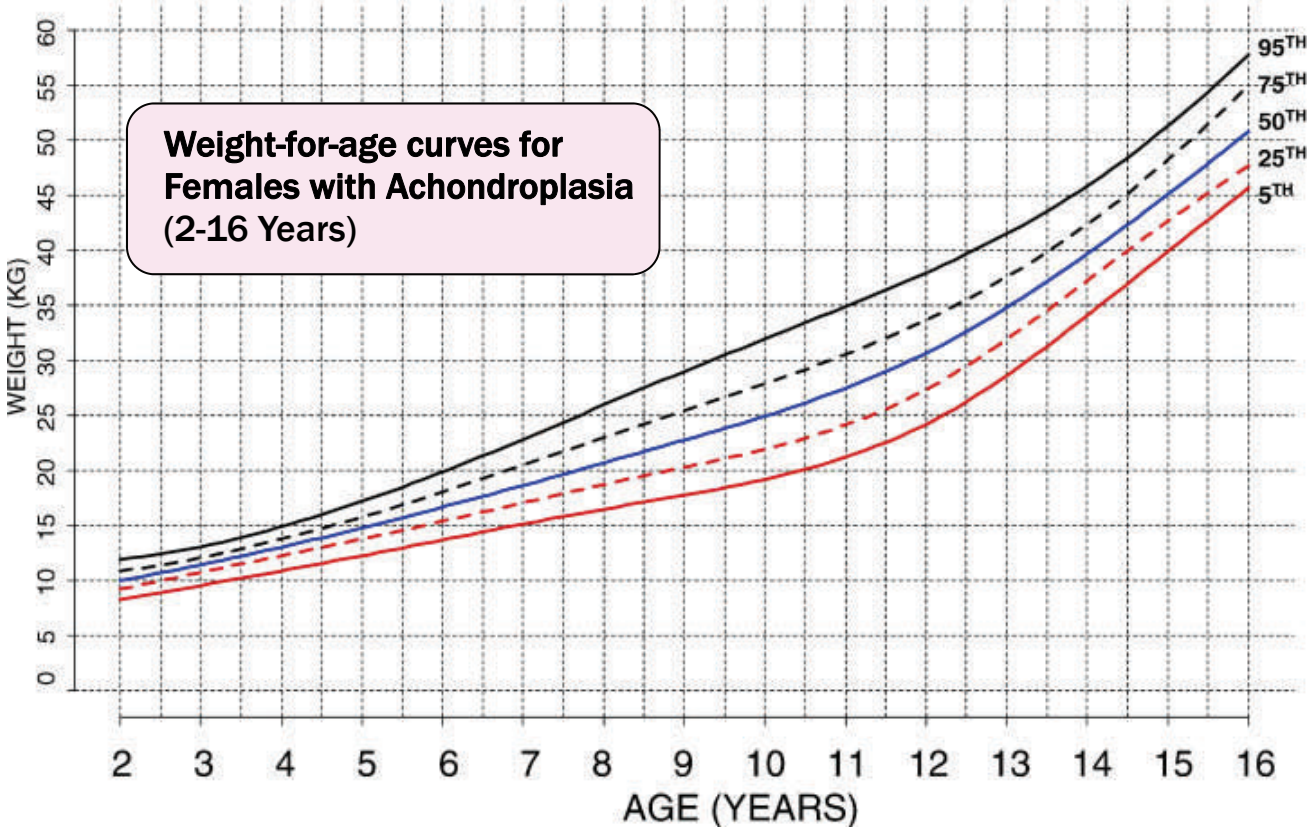
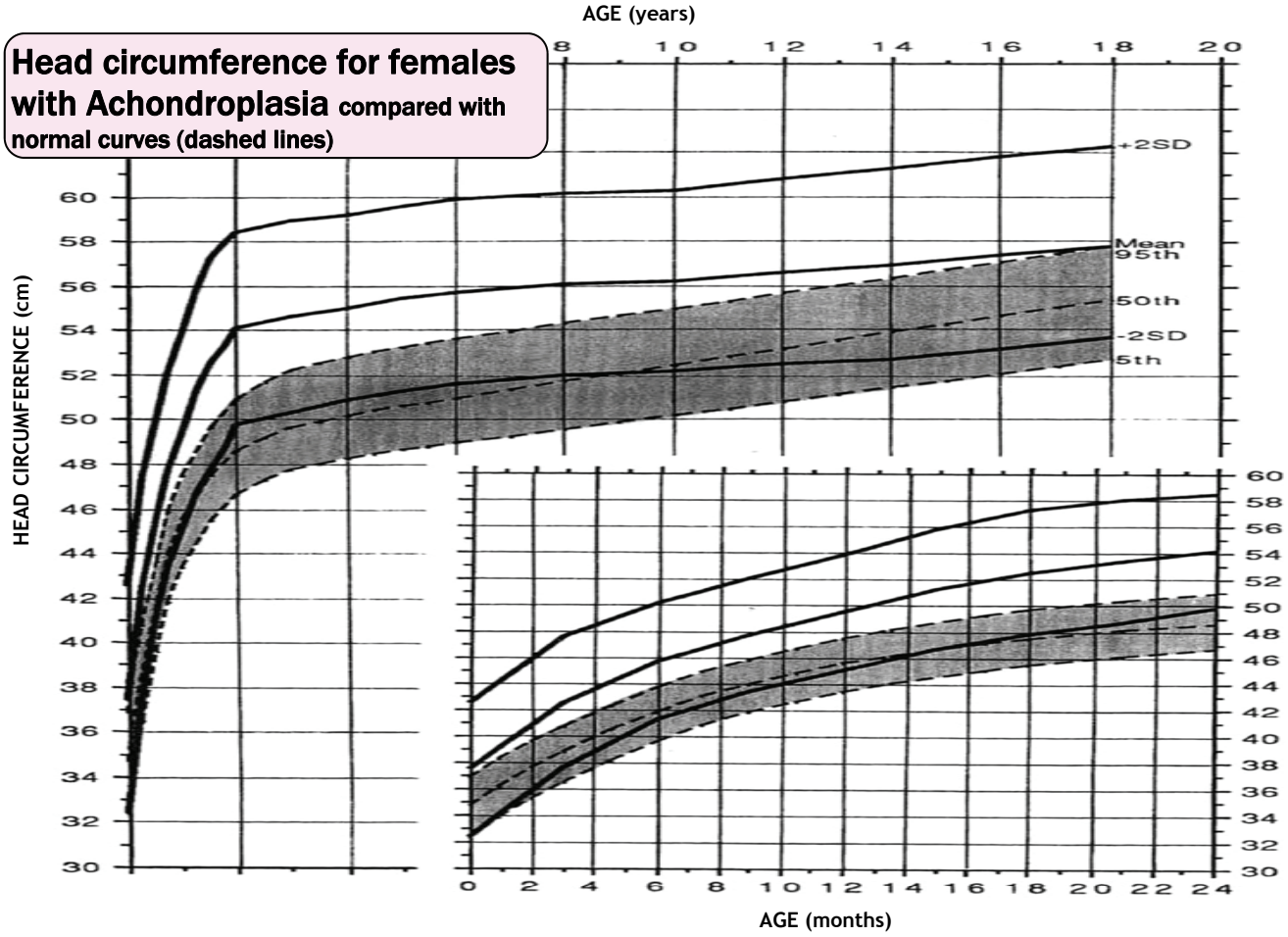






Head circumference for females with Achondroplasia compared with normal curves (dashed lines)



My Notes

Sheet Number

You can use this page for any other information you wish to remember or record. For example, use it to write down any questions you want to ask at your next appointment. As your doctor for more sheets if you need them.

Sources of Information

UK Restricted Growth Association
Telephone: 01935 841 364 Website: www.rgaonline.org.uk
The Restricted Growth Association is a non profit organization that provides information and support to improve the quality of life for persons of restricted growth, and promote the interests of people of restricted growth and their families. Gatherings of RGA members take place across the UK, centring on the Annual National Convention, also including Youth and Young adults Weekends, Family National Days and Regional Socials. New members are made very welcome whether they are mothers or fathers with a child of restricted growth, or a person of restricted growth themselves.

Contact A Family
Helpline: 0808 808 3555 Website: www.cafamily.org.uk
The Contact a Family website is for families who have a disabled child and those who work with them or are interested to find out more about their needs. Contact a Family is the only UK charity providing support and advice to parents whatever the medical condition of their child. They have information on over 1,000 rare syndromes and rare disorders and can often put families in touch with each other.

Internet
The internet contains a wealth of information about medical conditions and treatments and users need to be confident that the information they read on the internet is reliable and accurate. The Health on the Net Foundation website (**www.hon.ch)** has guidelines that help you make informed decisions about the quality of Web sites, and gives advice on how to search the internet for trustworthy health information. The Foundation has developed an ethical code of conduct (HONcode) and websites that publish health information can apply to display an HONcode seal/ badge, to show that they adhere to this code.