## **Consultation Guide**



			-	European Achondroplasia Forum
	ASSESSMENT	ACTION	~	REFERRAL
ALL LANGES	Do you have any symptoms/signs relating to <b>SPINAL STENOSIS</b> ?	<ul> <li>Neurological assessment</li> <li>Referral to MRI of the total spine (spinal stenosis can occur at multiple spine levels)</li> </ul>	то:	Neurologist (experienced in achondroplasia)
	<ul> <li>Ongoing back pain in combination with pain or discomfort radiating into the buttocks or legs, worsened by prolonged walking or standing</li> </ul>			Spine centre/specialist experienced in achondroplasia
	• Reduced sensation, tingling or numbness in arms or legs		IF:	Ongoing back pain in combination with neurological symptoms
	<ul> <li>Symptoms typically relieved by rest, lumbar flexion, or squatting</li> </ul>			· ·
	<ul> <li>Reduced walking distance/ increased need for walking aids</li> <li>Bladder and/or bowel dysfunction</li> </ul>		WFIEN:	As soon as possible to avoid spinal cord injury
	Do you have any symptoms/signs relating to obstructive <b>SLEEP APNOEA</b> ? • Excessive daytime sleepiness	<ul> <li>Overnight sleep study (ambulant or polysomnography)</li> </ul>	то:	Respiratory/sleep specialist (experienced in achondroplasia)
	<ul> <li>Not feeling rested after sleep</li> <li>Loud, disruptive snoring</li> <li>Breathing stops, choking, or gasping during sleep (observed)</li> </ul>		IF:	Excessive daytime sleepiness in combination with at least one of the other symptoms/signs
	Hypertension		WHEN:	As soon as possible
	BLOOD PRESSURE MONITORING	• Treatment as for the general population		
	SYMPTOMS OF HEARING LOSS	<ul> <li>Referral for a formal hearing assessment if hearing loss is suspected</li> </ul>	то:	ENT specialist
	WEIGHT MANAGEMENT	<ul> <li>Discuss the importance of an active and healthy lifestyle to prevent excessive weight gain</li> <li>Advice as for the general population</li> </ul>		
	PAIN	<ul> <li>If symptoms of pain, perform a formal pain assessment</li> </ul>		
A	NEED FOR ADAPTATIONS/AIDS	Referral if needs identified	то:	Occupational therapist
	GENETIC NATURE OF ACH	<ul><li>Discuss heritability and pregnancy (if needed)</li><li>Referral if needed</li></ul>	то:	Clinical geneticist or genetic counsellor
	PSYCHOSOCIAL HEALTH	• Evaluate psychosocial health. Refer if indicated	то:	Psychologist or psychosocial health worker