

ASSESSMENT	AT DIAGNOSIS	FOLLOW-UP FREQUENCY	AS NEEDED	PRE-ERT
Medical history	X	Every visit		
Physical examination	X	Every visit		X
Upper limb function • Standardized function test	X	Annually		X
Hips and lower extremities • Hips/pelvis: radiograph • Lower extremities: radiograph	X X		X X	
Spine/spinal cord compression • Plain radiograph • MRI • CT scan	X X	Every 1-3 years Annually	X	
Cardiac function • ECG • Echocardiogram • Heart rate	X X X	Every 1-3 years Every 2-3 years Annually	X X	
Respiratory function • Lung function • Breathing capacity • Ability to exercise • Overnight sleep study	X X X X	Annually Annually Annually Annually		X X X
Neurological function • Neurological exam	X	Every visit (minimally every 6 months)		X
Ophthalmological function • Vision assessment • Evaluate eye shape irregularities	X X		X X	
Hearing • Hearing assessment	X	Annually		
Dental evaluation • Evaluation of oral health by dentist	X	Annually		
Endurance • 6MWT • Timed 25-foot walk	X X	Annually Annually	X X	X X
Growth • Height and length • Weight • Head circumference (infants ≤3 years) • Pubertal stage (age 9 until mature)	X X X X	Every visit Every visit Every visit Every visit		X X X
Disease burden • Pain assessment • QoL questionnaire • Functional test • Activities of daily living questionnaire	X X X X	Every 6 months Annually Annually Annually		X X X X
Evaluation by physiotherapist	X	Annually	X	

*Adapted from the American Journal of Genetics International Guidelines for the Management and Treatment of Morquio A Syndrome.