Checklist for paediatricians and GPs.
Recommended follow-up of persons with osteogenesis imperfecta (OI) in Norway

Everyone who has OI should have a good working relationship with their General Practitioner (GP) . All children with OI should have a review with their paediatrician as soon as the diagnosis is made and make a plan for follow-up. Adults with OI should have a consultation with their GP with a review of symptoms and findings related to the diagnosis approximately once a year (adapted to individual needs). Detailed checklists for children and adults can be found below.

What is osteogenesis imperfecta (OI)?

OI, also called brittle bone disease, is a group of hereditary connective tissue diseases that primarily affect the skeleton, but other organs are also affected. OI is divided into subtypes with great variation in clinical findings and severity.

Common characteristics of OI are increased fracture tendency, deformities in the back, arms and legs, hypermobile joints, blue sclera of the eyes, decreased hearing, tooth changes and varying degrees of short stature. In addition, various internal organs such as lungs, heart and stomach / intestines may be affected. Despite the medical challenges, people with OI live active and good lives.

Read more about medical aspects of osteogenesis imperfecta (OI) (in Norwegian) – at TRS resource centre for rare disorders.

What should be followed up regularly?

Children with OI

Age, severity and the combination of symptoms and findings determine what follow-up and treatment each child with OI will need. There is no curative treatment for OI. The goal of the follow-up is therefore to contribute to the best possible quality of life, increased mobility and functional independence.

Treatment is aimed at improving bone strength, reducing fracture risk, limiting pain, correcting deformities, and preventing long-term complications.

All children with OI should have a review with their paediatrician as soon as the diagnosis is made and make a plan for follow-up. Children with clinically moderate and severe OI should be monitored regularly by a paediatrician, paediatric orthopaedic surgeon and physiotherapist (possibly at an OI – clinic), frequency is agreed individually.

See checklist for follow-up of children next page.

Checklist for follow-up of children with OI

What should be investigated?	***Recommended assessments***	***How often?***	***Comment***
Skeleton	X-ray total skeleton on suspicion of OI Common changes: Curved bones, foot misalignment (flat feet), increased spine curvature (kyphosis/scoliosis), wormian bones in the skull. Fractures, deformities and bone density measurements determine the indication for treatment with bisphosphonates		
Fracture	Ask if there have been new fractures since the last consultation. Low threshold for X-rays on suspicion of fractures, but fresh OI fractures can be difficult to detect.		Children with OI fractures should have easy access to hospital with an orthopedic surgeon
Bone density measurement	Norway unfortunately do not have access to bone density measurements for the very youngest	First time at 4 years of age	
Hearing	Refer to audiometry, refer to ENT specialist in case of symptoms	First check-up at 4 years of age, then approximately every 3 years or in case of new symptoms	Approximately 50% of all people with OI have hearing loss
View	Refer for eye examination - refer to an ophthalmologist if symptoms occur	First check-up before school starts, then approximately every 3 years or in case of new symptoms.	Follow-up depending on findings
Teeth	Follow separate guidelines for follow-up of teeth and oral cavity at OI, from the Norwegian Centre for Oral Health in Rare Disorders (TAKO Centre) (in Norwegian)	First examination at 6 – 8 months of age when teeth first arrive	
Function	Monitor the child's development, especially new deformities in the spine, feet and arms/legs. Refer to pediatric orthopedic surgeon if findings	Annually/on demand	Change since last?
Pain	Simple pain anamnesis – VAS scale with faces	Annually/on demand	Change since last?
Blood tests	Checking total calcium and vitamin D Other blood tests if needed	Annually/on demand	Give supplements at too low values
Nutrition	Review of diet – is the child getting enough calcium and vitamin D? Note! Overweight in children who are immobile. Note! Constipation is not uncommon	Annually/on demand	Provide info about proper and varied diet
Transition from child to adult (transition)	When the person turns 14 - 16 years old, they should be offered counselling about switching to adult follow-up		

Adults with OI

Consultations with a GP should include conversation about, and examination of, the following issues:

What should be investigated?	***Recommended assessments***	***How often?***	***Comment***
Blood tests	Checking total calcium and vitamin D Other blood tests if needed	Annual	Start treatment at too low values
Skeleton	Refer to bone density measurement (DXA) In osteopenia / osteoporosis - refer to the endocrinologist to assess the indication for treatment with bisphosphonates. Attention to increasing spinal deformity (kyphosis/scoliosis)	Every 5 years unless otherwise agreed with specialist or changes have occurred	Attention to new fractures, especially in the spine, and increasing skeletal pain
Fracture	Ask if there have been new fractures since the last consultation. OI fractures can be difficult to detect	Refer to orthopedic surgeon v/deformities, fractures that do not heal, increasing pain	
Hearing	Refer to audiometry, refer to ENT doctor in case of symptoms	Every 3 years or in case of new symptoms	Approximately 50% of all people with OI have hearing loss
View	Simple eye test – refer for eye examination - refer to ophthalmologist if symptoms occur	Every 3 years or in case of new symptoms	Measuring eye pressure can give false values
Teeth	Follow separate guidelines for follow-up of teeth and oral cavity at OI, from the Norwegian Centre for Oral Health in Rare Disorders (TAKO Centre) (in Norwegian)		
Lungs	Spirometry, listening to lung sounds (auscultation) - asking about heavy breathing, morning fatigue. Refer to pulmonary doctor	First examination at 25-30 years of age + when symptoms	
Cardiovascular	Listening to heart sounds (auscultation), ECG, BT – refer to cardiologist	First examination at 25-30 years of age + for symptoms	
Function	Conversation about function – experiencing loss of function? Physical activity? Possible mapping of function – simple ADL status	Annually/on demand	Change since last? Need for rehabilitation?
Joint Status	Hypermobile joints?	Annually/on demand	
Pain	Simple anamnesis / pain assessment	Annually/on demand	Change since last?
Nutrition	Review of diet – important that calcium and vitamin D are covered through natural sources. Note! Constipation is not uncommon	Annually/on demand	Constipation can also be caused by skeletal change (protrucio) of the hip
Other	Have a low threshold for referral in the event of new symptoms/findings	Annually/on demand	