Condition Insight Report (CIR)

Brittle Bone (Osteogenesis Imperfecta)

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Completed in collaboration with Brittle Bone

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Overview

What is the condition usually called / any abbreviations used?

Brittle Bone Disease or Osteogenesis Imperfecta (OI)

OI presents clinically in a very similar way to Ehlers Danlos Syndrome (EDS) but with the added issues of fractures and other symptoms.

Brief overview of the condition

OI is caused by a genetic mutation that affects the production of collagen. In OI the collagen may be of poor quality, or there may just not be enough to support the mineral structure of the bones. This makes the bones weak and fragile and results in the bones being liable to fracture at any time even without trauma (i.e. waking up with a cracked rib). As the composition of collagen in the bones is not correct, even when there are no fractures, there will be other problems connected to the condition such as: ligaments that stretch more easily and joint hypermobility which can significantly affect quality of life, as it results in fatigue within large muscle groups. As a result mobility and performance of ordinary tasks of everyday living are impaired. OI tends to lead to individuals requiring the use of walking sticks, crutches, walking frames and wheelchairs for daily mobility. Some individuals will walk very short distances around their home, however throughout the day they will become more fatigued and require a wheelchair or walking aid, enabling them to continue with their daily activities in a modified way.

What is the generally preferred term for someone with this condition?

Living with OI or they **may refer to themselves as an OIer**

Presenting Symptoms

Can include, but not limited to:

- Fractures
- Hypermobility
- Propensity to fall
- Soft tissue injury
- Bruising with little or no effort
- Dislocation / subluxation with little effort
- Short stature
- Deafness / hearing loss
- Fatigue
- Chronic pain, including chronic bone pain
- Poor sleep quality / insomnia
- Poor grip
- Reduced power
- Clumsiness
- Poor manual dexterity
- Joint laxity
- Mobility issues
- Curvature of the bones
- Scoliosis / kyphosis (curvature of spine)
- Dentinogenesis imperfecta (involvement of teeth)
- Mental health issues; anxiety, depression, agoraphobia, PTSD
- Reduced pulmonary (lung) function causing breathlessness with routine activities
- Heart valve problems
- Issues associated with high levels of pain and high dose pain medication - brain fog, poor concentration and focus
- Weakening of the vessels, increased frequency of nosebleeds etc.

Fluctuations

The condition can deteriorate with injuries but generally, over time the cumulative effects increase, mobility reduces and symptoms increase. In adulthood, even in mild forms of the condition the impact on daily living and mobility can be substantial and permanent.

Think about exploring things like:

- Frequency of fractures, hospital admissions, management plans
- What minimal exertion or activity can cause fractures?
- **How has the condition changed over time?** e.g. the last 12 months
- **Changes in treatment** or discussed future treatment options recently and what the expected outcome is

For pain, fatigue and breathlessness:

- **What level?**
  Can they quantify this, do they use a scale and can they describe it?
  Can they discuss this in this way over what might be a 'typical' day for them?
- **Difference between symptoms at rest and** what occurs **during exertion. What level of exertion causes a change**, how long any changes in presentation then can last and impact this has?

Reliability

What specific areas should be covered to ensure a complete, reflective report?

SAFETY	TIMELY	ACCEPTABLE STANDARD	REPEATEDLY
Do they have any symptoms which could cause a safety consideration?	For any activities where restriction is reported how long does it take them to complete these activities? Has how long it takes them changed over time?	How have they adapted to completing tasks over time – is this different to what might be considered 'normal'?	Are they able to repeat a task as often as required? Is this the same every day?
Multiple fractures over time leads to deformity in long bones and spine, this deformity makes functional tasks more difficult and can result in chronic pain. Due to the high risk of fracture safety needs to be considered in all appropriate areas.	All self-care tasks and mobility is likely to be done at least 2-3 times slower than the average person, with pain present throughout, fatigue and for some breathlessness. Determining how this is managed for each individual must be explored, as whilst aids can be available they cannot always, if at all, mitigate their risks of damage from the prolonged exertion.	Some adult OIers, in order to be independent, require carers/assistance dogs/support workers in order to maintain themselves and their homes. Due to there being various types of OI from mild to severe, this will vary for each claimant. Even in the mildest forms it is rare for daily living not to be affected by the condition. If they have other conditions over and above OI then the impact can be magnified.	The ability of the OI community to withstand high levels of pain can mask the true extent of the problem. Used to putting up with a constant pain as a matter of course. Pain management, mental wellbeing and fatigue are the "hidden disability". Hypermobility is always prevalent but seems to be unacknowledged in comparison to fractures. Also the effect of heavy-duty pain relief on function and cognitive ability.

Sensitivities

What areas might they find difficult to mention or perhaps understate the impact of?

People naturally want to think about what they can do, and not about what they can't do, so it is difficult for someone to change their mind set when being questioned.

Young people may still be trying to acknowledge that they have a disability and want to be treated as normal and like their peers. They often don't realise until they are going to University or, entering the workplace what adaptations are actually required or what additional help they have been receiving from parents prior to leaving home.

Others may not start having symptoms until they reach their 40's and find it difficult to admit they are struggling with daily tasks.

Watch **OI awareness video**
HERE

Customer Care

How is it best to ask about any sensitive topics and what are the common courtesies?

In general

- **Be personable, friendly and honest.**
- **Do no refer to it as a disease – it is a disorder you are born with and has no cure.**
- **Please don't tell them you hope they will get/feel better.**
- **Be mindful that they may become emotional. Acknowledge you appreciate certain topics may be hard to discuss and offer a break as required.**
- **Acknowledge that the claimant is the expert in their own lived experience.**
- **Ask if they had to prepare for today - increased medication, bring advocate/support, did they curtail activity prior etc.**

During face to face interactions

- **Face the client, ensure they can see your mouth as likely hearing loss.**
- **Never assume they need assistance, or they won't want any. Just ask.**
- **Ensure chair is comfortable and allow them to feel comfortable to move, change position or rise as needed.**
- **Due to how prone to fracture Olers might be, it may be inappropriate to ask them to complete an MSK**

Functional Impact

A brief summary of the functional impact those living with this condition may experience

Activity 1: Preparing food

Poor manual dexterity, clumsiness, reduced power and grip, difficulty standing or maintaining prolonged posture, difficulty carrying objects, particularly for those individuals who have short limbs. Individuals with OI are likely to need an adapted kitchen e.g. lower worktops/appliances, perch stools, step stools, adapted utensils/ cutlery, lightweight pots/pans and crockery, kitchen trollies. Other useful items would be; automatic tin openers, jar openers, choppers, anti-slip mats, sip cups/straw cups, kettle tippers /light kettles. The effort of cooking would cause fatigue and pain. Increased likelihood of injury when cooking e.g. cuts, scalds and burns. Assistance or another person doing the task entirely would be likely.

Remember in PIP…

Can someone prepare and cook a simple meal for one? How long can they stand in the kitchen? Can they chop and peel vegetables? Can they lift pans? Does completing this activity increase breathlessness or fatigue? How do they feel after the activity? How long does it take to recover? Do they have pain or are they at risk? Are they motivated?

Activity 2: Taking nutrition

Holding cutlery, cups etc. can be difficult. Use of straws, adapted cutlery/crockery, picking up food, avoidance of food that requires cutlery, need for certain crockery to allow for need for lightweight and easy grip. Difficulty chewing due to either jaw malalignments or dentinogenesis imperfecta (brittle teeth). Slow to eat. Prone to spillage and especially problematic if the food/drink is hot. May avoid such things and will choose soft food or food that can be picked up rather than use cutlery. Drinks need to be in lightweight vessel that can be held with both hands. Straws are often used by people who have OI due to reach issues, or when spending long times laid down when fractured.

Remember in PIP…

Can they chew, swallow and bring food to their mouth? Can they physically cut food? Do they have any aids to assist such as adapted cutlery? Can they eat solid food? Do they have any risk of choking? Can they complete this activity in a timely manner?
Do they need prompting to eat?

Activity 3: Managing therapy and monitoring a health condition

Therapies are a daily event. This can be passive and active. Not always aware of limitations so can “crash” if body is pushed beyond limit. May need assistance with placement of devices e.g. TENS machine, heat packs etc. due to reduced range of motion. Therapies can include physiotherapy, hydrotherapy, massage and even swimming etc. However due to propensity to fall and difficulties with other aspects such as changing etc. then supervision and assistance will be likely. Likely been to physios, consultants and OTs throughout life so unless at a point of injury or operation they do not have regular contact with health care professionals. OIers tend to be very adept at self-treatment and are able to bind and care for injuries without need for A&E/GP intervention. They may have fractures that have not been logged in medical notes due to this.

Remember in PIP...

Therapy is only considered within the scope of the activity if received in the home environment. If they do have therapy at home, do they require assistance? If so, why and how long does this take?
Regarding their medication, can they manage this themselves? Can they administer their own medication? Can they remember to take it and physically remove it from packaging? Do they need prompting?

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Activity 4: Washing and bathing

Difficulties include getting in/out of the bath or shower. Many rely upon baths as well as showers as submerging in hot water can ease joints so is a therapy as well as for cleansing but has increased risk of slipping and further injury. Problems with reaching all required body parts especially back, head and hair, and feet. This task alone can be exhausting and is likely done in stages due to fatigue and pain caused. May avoid washing as often as needed for this reason. These difficulties increase with fractures and injury. Bathroom adaptations include wet rooms/walk in showers, lower baths, hoists, slide boards, stools to reach toilet/basin/bath, derby steps, anti-slip mats/floors, grab rails, toilet frames, raised toilet seats, easy to use bathroom products.

Remember in PIP…

You must explore how someone is managing the activity. How do they get in and out of the bath? Do they have any aids? Can they wash their entire body? How long does it take and how do they feel after? Does this activity increase their symptoms? How long does it take to recover? Do they need assistance with upper or lower body? Are they at risk of falls? Do they have adaptations?

Activity 5: Managing toileting needs and incontinence

Depending upon the severity of OI an individual may require a carer to assist with these tasks or they may be able to self-manage with some difficulty and/or pain experienced. Difficulty wiping/reaching - may use wipers or specialised equipment. Difficulty using standard toilets due to short stature, joint deformity, pain and fatigue. Toilet frames, easy flush toilets, moist wipes, bidet attachments and auto dryers are among the aids that can be utilised. Outside the home environment toileting can be very problematic so incontinence aids are likely.

Remember in PIP…

You must explore how someone gets onto and off the toilet and how they clean themselves? Does doing this make them more breathless? We do not consider ability to mobilise to the toilet. Also explore if someone has incontinence on majority of days and how they manage this.

Activity 6: Dressing and undressing

This activity is slow and laboured due to pain, reduced range of movement and fatigue from the effort involved. Clothing may often need to be altered or hard to source due to short limb length, barrel chest, and deforming caused by fractures, bowing and such. Fastenings can be especially difficult. Zips, buttons, hook and eye can make all aspects of dressing difficult. Bras either need to be front fastening or loose. Even over the head styles can cause injury due to joint laxity when trying to dress. It is likely they cannot reach upper parts, lower parts, shoes, socks/tights and reach around to bra's and zippers, will likely need assistance or aids, if available. Dressing aids can include shoehorns, tight/sock pullers, zip pullers, velcro fasteners, larger zip pulls and buttons, elastic laces/non-laced footwear. Supports/braces/casts/orthotics also need to be accommodated so baggy clothing is likely.

Remember in PIP…

You must explore someone's ability to dress and undress within the scope of STAR. How long does it take them? Can they dress upper and lower body? Does this increase symptoms of fatigue or pain? Do they require assistance or use any aids? Can they dress reliably whilst seated?

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Activity 7: Communicating verbally

50% of OIers have deafness/hearing loss. Aids are not always a solution to this although many do wear hearing aids. Communication can be problematic especially if dealing with officialdom or in crowded, noisy, social settings. Please check for understanding on questioning as some may lip read or use sign language. Subtitles are frequently used for TV/Film.

Remember in PIP...

Can someone express and understand both basic and complex verbal information? Do they utilise bilateral aids? If they cannot use aids, do they lip read? What can they hear? How do they communicate?

Activity 8: Reading and understanding signs and symbols

While OI does not specifically cause an impairment of cognitive function, the levels of medication a person may be on could affect their ability to comprehend. High dosages of opiates and similar analgesics coupled with poor sleep quality can cause symptoms such as brain fog, delayed reactions and poor concentration and focus feature typically. Any fixed positioning can be painful so must regularly change position so reading/holding papers or books can be an issue. Book rests/iPad/phone rests may be used as well as dictation/speakers or voice-activation.

Remember in PIP…

Can someone read and understand basic and complex written information? What do they read? Can they read a text message or a book? Did they gain any qualifications in education?

Activity 9: Engaging with others face to face

The energy needed to conduct day-to-day life may mean that socialising is harder. May require both physical and mental assistance. Hearing issues can be problematic in a social setting. The consequence of engaging with others can result in days of bed rest so they may avoid socialising because of this. Social stigma can exacerbate anxiety and depression when abuse has been encountered.

Remember in PIP…

Remember to explore how they feel when engaging with both familiar and unfamiliar people. Do they have anxiety? If they do, who can support them to engage and why is this? Do they isolate themselves due to their condition?

Functional Impact

A brief summary of the functional impact those living with this condition may experience

Activity 10: Budgeting

While OI does not specifically cause an impairment of cognitive function, the levels of medication a person may be on will affect their ability to comprehend and make budgeting decisions. High dosages of opiates and similar analgesics coupled with poor sleep quality can cause symptoms such as brain fog, delayed reactions and poor concentration and focus feature typically. Whilst not covered in A10 the physical components of handling money can provide further detail about their level of function, such as accessing cashpoints etc.

Remember in PIP…

You must explore someone’s ability to manage both basic and complex budgeting. This would include how they manage their household budgets. How do they plan for future purchases? Would they understand change to expect in a small transaction? Is this area impacted by low mood or any cognitive impairments?

Activity 11: Planning and following a journey

Anxiety and fear of being in certain environments, accessibility issues, not being able to travel for long distances or not having a carer to take them can lead to isolation. Depending on the length of journey and accessibility, the journey may need to be planned well in advance.

Remember in PIP…

Physical restrictions are not considered within the scope of the activity. Consider someone's ability to plan and follow a journey in relation to potential sensory or cognitive issues or the presence of OPD. How do they manage both familiar and unfamiliar journeys? Can they plan a journey? Could they manage a diversion? If anxiety is reported, does this meet the threshold of OPD? If so, could they leave the home on the majority of days with support?

Activity 12: Moving around

Some can weight bear, some can’t. For those that can weight bear the risk is damage, deterioration when on foot and weighing up when assistive equipment and care is needed, fatigue/pain throughout the day also means equipment likely needed such as a wheelchair, later in the day. Many use sticks, crutches, walking frames or wheelchairs to move. The ability to self-propel in a chair does not always denote ability. Repeated movements like this can mean damage elsewhere in the body so upper limb impairment can result from overcompensation. Self-propelling a heavy wheelchair over a long period of time is extremely fatiguing for individuals with OI. Mobilisation is slow, laboured and rarely without pain. They may be able to go out but know they will need to take the next few days to recover from their fatigue. They may also run the risk of being knocked if using public transport and breaking bones. Fractures/injury/hypermobility and operations can mean that supports, splints, orthotics, casts, support hosiery and similar need to be worn.

Remember in PIP...

Firstly, establish when someone is walking, do they require an aid? If so, what aid? Was this prescribed or self purchased?
How far can they walk and how long does this take? How does walking make them feel? Does pain/fatigue increase each time they walk? When they walk a second time, does pace reduce or distance reduce? If they experience fatigue, how do they feel later in the day? How many times could they repeat the distance they walk? How long does it take to recover from mobilising?
Lived examples are also extremely beneficial. How do they manage up and down the stairs? How do they manage with chores? How do they manage their shopping? How many aisles can they walk and how long does it take? If they go to their GP surgery or hospital, how far is it from the car? How long does it take them to get to the required department and how far is this?

Additional reading or other resources

EXTERNAL

https://brittlebone.org/what-is-oi/about-oi/

https://brittlebone.org/support/information-resources/

https://www.youtube.com/channel/UCJEDijvQWV-BuJi5HFKsv-w/videos

INTERNAL

- Desktop Aid – Fatigue, Breathlessness, Activity 12

Version control