

# Condition Insight Report (CIR)

## Fibrous Dysplasia / McCune Albright Syndrome

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Completed in collaboration with  
FDSSUK

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# Overview

## What is the condition usually called / any abbreviations used?

Fibrous Dysplasia (FD) (Polyostotic/Monostotic) / McCune Albright Syndrome (MAS)

Other terms are: MFD, PFD and CFD (craniofacial FD)

## Brief overview of the condition

FD and MAS are rare conditions caused by a genetic mutation in the GNAS1 gene at the time of conception, affecting any number of bones in the body (more commonly the skull and long bones) as well as other organs of the body such as hormone glands. It is not hereditary.

Fibrous Dysplasia is a progressive bone disorder and refers to bone which is replaced by fibrous connected tissue. This abnormal fibrous tissue weakens the bones, making them very fragile and prone to spontaneous fracture, break or become deformed. Fibrous Dysplasia bone does not heal in the same way as normal bone after a fracture, often leading to severe deformity, nerve damage and chronic pain. The abnormal bone can also become very painful and put pressure on the nerves and if cranial, can lead to visual and hearing problems. If the back becomes too twisted it can restrict breathing.

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

Living with FD, MAS.

# Presenting Symptoms

Fibrous Dysplasia is a progressive disorder which can lead to an **abnormal gait**, long bones in the legs often have **frequent fractures** due to weight-bearing (through walking and standing), which surgery can help but can cause its own complications, bone deformities such as **severe Scoliosis** (which is quite rare), and femur bones often develop a 'shepherds crook' leading to **leg length discrepancy**. Pain will continue to get worse as the patient gets older.

### Primary symptoms include:

- **Fractured/broken bones**
- **Bone Cysts**
- **Short Stature**
- Dental issues
- Thyroid problems
- Pituitary problems
- Severe Vitamin D deficiency
- **Visual and hearing impairments**
- Adrenal gland problems - rare in adults
- Abnormal levels of Phosphate in the blood
- Sinus issues - if the skull bones are affected
- Changes in appearance - when the skull bones are affected
- **Chronic pain** - Which is difficult to treat with current treatments
- Precocious Puberty, Ovarian Cysts and other hormonal issues
- Café-au-Lait skin pigmentation and Café au Lait spots (like birthmarks)
- **Abnormalities of the heart, pancreatitis, liver and abnormalities of the brain**
- **Reduced power/limited mobility** - due to bone deformities and multiple surgeries
- **Nerve compression** - in particular the optic nerve and ear canal, although this is rare
- **Mazabraud syndrome** - lumps occurring in the muscles that can restrict further movement and mobility
- Osteonecrosis of the jaw (ONJ) - this is extremely rare but can occur when using Bisphosphonates

### Secondary Conditions include:

- **Fatigue**
- Gastro issues - due to medications
- **Nerve pain** - due to multiple surgeries
- **Brain fog/poor concentration** - due to heavy medications
- **Psychological/mental health issues** - due to changes in appearance, anxiety, depression, isolation

McCune Albright Syndrome is characterised by; Polyostotic Fibrous Dysplasia, Café-au-Lait skin pigmentation and Endocrine Disease.

There is no cure for the disorder, it is lifelong, with no new treatments and there are lots of secondary conditions that will occur throughout a patients life. Each patient's condition is different, depending on the amount of bones and hormones affected in the body, and they will have their own individual needs. Not all patients are diagnosed in childhood, some are diagnosed in adulthood. An operation to fix a break or a fracture will not cure the disease. Patients can have several pains in different parts of the body at the same time. It is a variable disease and patients will have good days and bad days with frequent flare ups.

# Fluctuations



Pain flare ups are very common. Patients have good days and bad days.

Think about exploring things like:

- **How has the condition been over the past 12 months?**  
What changes have they seen, over what period of time did they experience any deterioration, what has this meant for their functional ability now compared to previous?
- **Explore the extent of any frequent fractures**, and extent of hospital admissions.
- **What extent of any mental health condition(s)?**  
Symptoms present at all times or triggered, how does it impact their ability to complete tasks?
- **What level of pain/fatigue?**  
Can they quantify this, do they use a scale and can they describe it? What level of severity? Can they discuss this in this way over what might be a 'typical' day for them?
- **Triggers?**  
Whilst exertion is a main trigger what amount of exertion is enough to cause further limitation? How are triggers managed?

# Reliability

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

**S**AFETY



**T**IMELY



**A**CCCEPTABLE  
STANDARD



**R**EPEATEDLY



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?

Depending on the reported symptoms, where it is appropriate cover how risks are mitigated and if there is a history of incidents within activities 1, 2, 3, 4, 5, 9, 11 and 12 to ensure you have addressed the needs of the individual claimant.

Safety considerations can be a result from physical and psychological symptoms.

Pain, fatigue, muscle weakness / bone deformities can all affect time frames of how long it takes an individual to complete a task. This will vary for each individual, but there are no specific time frames outlined for what is reasonable.

## Ensure to ask:

- How long it takes to complete tasks such as 1,4,5,6 and 12.
- Why they feel it takes them this long?
- Where they report having any breaks in task, how long are these breaks and how frequent?

As FD can cause various impacts how the individual has adapted to e.g. their leg discrepancy will become their 'normal'. This may mean they do not raise this during superficial questioning.

Exploring **HOW** they complete the task in depth can help to draw out any compensation strategies which can then be explored further.

Ask about the pain and fatigue experienced whilst carrying out activities and then post the activity.

As pain and fatigue can vary, how the individual is on any given day is not how they are going to be all the time. You need to try and establish what is the majority of times for them.

For example, someone with chronic pain, they may be able to complete an action but then be unable to do anything following this or repeat it again in the same day.

# Sensitivities

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

- Extent of changes over the course of their condition
- How far they have adapted tasks
- Their emotional state and other hidden symptoms not easily identified e.g. fear of falling, isolation
- What it is like during a bad phase of their FD/MAS.
- Chronic Pain / discomfort & different types of pain and impact on their ability to concentrate and energy levels.
- Alternative treatments
- Adaptations
- Weight gain
- Toileting and associated incontinence

Watch **Jamie** talk about her McCune-Albright Syndrome **[HERE](#)**

Watch **Kay** talk about her Fibrous Dysplasia **[HERE](#)**

# Customer Care

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

In general

- Do not ask too many questions at once
- Present one question at a time and give time for a response before asking another question
- Involve any companions
- Be clear you are going to assess how their condition impacts on their daily lives, not the condition itself
- Try and recognise, if possible, when the claimant is deteriorating and becoming more physically and cognitively distressed – ask if they want to stop, take a break etc.



During face to face interactions

- Signpost individuals to toilets and exits
- Support by holding open any heavy doors
- Confirm they can get up and move around should they need to
- Access to a comfortable chair where possible
- Check whether any sensitivity to light/noise and make adjustments to the environment if possible
- Maintain eye contact
- Think twice before asking them to complete an MSK or shaking the individuals hand



# Functional Impact

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

## Activity 1: Preparing food

They may use aids or require assistance for lifting pans or bending to remove items from cupboards. Some patients who use wheelchairs may need to have their kitchen adapted so that the cupboards and oven are low level or eye level. Other patients are unable to stand for long periods of time, bend or reach up into cupboards. Due to associated mental health conditions some may require prompting. Physical impact such as changes in respiratory function, pain, affected vision, sensitivity to fractures can hugely impact this activity where some may require full assistance depending on the severity.

### Remember in PIP...

Can they safely cook a meal? Are there any risks in the kitchen as a result of their condition? Can they stand to prepare a meal? If so, how long for? Can they chop vegetables? How do they feel during and after the activity? Do they require prompting to cook? Can they transfer from their wheelchair or do they require assistance to cook?

## Activity 2: Taking nutrition

Patients who have FD in the skull bones often have problems chewing food. Due to associated mental health conditions some may require prompting. Physical impact such as affected vision, extent of scoliosis can impact this activity where some may require assistance to cut up food and/or take it to their mouth.

### Remember in PIP...

You must ensure you explore if someone is adequately nourished. Can they cut their food, bring it their mouth, chew and swallow? Do they require an adapted diet or blended food? Are they at risk of choking? Do they need prompting to eat? If so, is this throughout the duration of the meal?

## Activity 3: Managing therapy and monitoring a health condition

Treatments and therapy are often temporary but there may be regular medication such as regular Bisphosphonate infusions or analgesia which individuals need support to take. There are no effective or new treatments available. Most people have to manage their own pain and there are a lack of specialist Dr's. Patients also have to manage their own mental health and wellbeing.

Alternative treatments they may use for example; heat pads/ tens machine/ ice packs/ massage/ physio/ hydrotherapy/ cryotherapy.

### Remember in PIP...

If someone is having therapy in the home, do they require assistance? If so, how long does this take? Is this for the majority of the year? Also, if they are taking medication, can they self manage this? Can they get it out of the packets and self-administer? Do they require prompting to manage their medication? Do they require assistance to apply things such as a tens machine?

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## Activity 4: Washing and Bathing

Sufferers may have issues getting in or out of a bath, they may require the use of a wet room/walk in shower, assistance due to lack of mobility in long arm bones, or lack of strength or balance in the legs. Those with severe deformity or wheelchair users may require a hoist to get in and out of the bath. Slip mats, grab rails, frames, assistance and/or supervision may also be needed. There must be sensible flooring in the bathroom to prevent falls. Associated mental health conditions may also impact their ability. This can take some extra time and it can be painful.

### Remember in PIP...

Can they wash and bathe majority of days? Can they reach all areas of the body? How long does it take them? Do they sit down, if so, why? Can they get in and out of an un-adapted bath? Can they overcome restrictions with aids, or do they require assistance? Do they experience pain or increased fatigue after the activity? Do they lack motivation to wash? Have they had falls? Are there any risks?

## Activity 5: Managing toileting needs and incontinence

Extent of scoliosis can hugely affect their ability to get on and off the toilet with considerations as to whether they reach to manage hygiene and how they have been affected by previous fractures and ongoing pain. Patients may need to use mobility aids, or if wheelchair bound may need assistance. Some women may need to use medication to control their menstrual cycle.

### Remember in PIP...

**Management of a menstrual cycle is not considered within the scope of the activity.**  
If someone reports incontinence, how are they managing this? Can they reliably use aids or do they require assistance? Is incontinence on the majority of days?  
  
Also, how do they physically manage toilet needs including sitting on the toilet, standing from the toilet and cleaning themselves?

## Activity 6: Dressing and undressing

Due to associated mental health conditions some may require prompting. Physical impact such as changes in respiratory function, pain, affected vision, sensitivity to fractures can hugely impact, restrictive and tight-fitting clothes can cause pain or discomfort. Alterations may be necessary due to deformity. Fastening of shoes may be difficult due to mobility and they may be impractical. This activity where some may require full assistance depending on the severity.

### Remember in PIP...

Being seated is considered acceptable. Does being seated improve their ability to dress? Remember to explore how they do this. They may report they can dress their body, but is this in an acceptable manner? How long does it take? How do they feel after? Do they require assistance due to the severity of their symptoms?



# Functional Impact

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## Activity 7: Communicating Verbally

Skull involvement can cause hearing impairment and vision loss, although this is very rare. Aids may not always be possible due to cranial deformity. The jaw and teeth can become affected too causing difficulty talking. Confidence can be an issue as people do not always understand that there is anything wrong if a patient looks normal to them, the condition can be invisible in some people. Also, confidence can also be difficult if there is a noticeable change in the facial bones and people stare. Heavy medication and fatigue can make communication and concentration difficult.

### Remember in PIP...

The scope of the activity is someone's ability to both express **and** understand verbal information. Can they understand what they have been told? Can they hear? Do they require bilateral aids? If so, are they effective? If not, how do they communicate? Do they need to lip read?

## Activity 8: Reading and understanding signs and symbols

If vision is impaired due to skull involvement, it can cause pain both around the eyes and the skull, difficulty looking at text or screens and may become light sensitive.

### Remember in PIP...

This is the ability not only to read written information, but also understand it. Do they require aids other than spectacles? Could they read a text message? How did they manage reading in education?  
**Within the scope of PIP complex written information is more than one sentence.**

## Activity 9: Engaging with others face to face

Feeling socially unaccepted, anxiety, dealing with chronic pain/headaches, deafness and social stigma of appearance or disability and the presumption of 'can't do' rather than 'how can' will also lead to exclusion and exacerbate anxiety, exclusion and segregation leading to depression. Loud noises and loud social environments causing intense skull pain. Sometimes if feeling anxious or unwell on the day of the event, sufferers may often cancel an appointment at short notice and people do not understand this. Relationships are also problematic as people do not understand the pain.

### Remember in PIP...

You must explore who the person can engage with on a regular basis. How do they feel engaging with unfamiliar people? Does this increase anxiety? If so, how is this managed? Do they require support? Is the support from someone specific? If it is, what do they do that nobody else can do?

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## Activity 10: Budgeting

Consider any impact of any associated mental health conditions on their ability to manage budgeting.

### Remember in PIP...

Can they manage both basic and complex budgeting? Would they understand what change to expect in a shop? Can they plan for future budgeting? Could they manage a household bill? Do they need support with any areas of budgeting? Are they motivated to manage their finances?

## Activity 11: Planning and following a journey

Sufferers can find it difficult and upsetting when coping with change i.e. if a bus is cancelled or if the train is too busy to board, extra time must be given so they can travel safely. For those who have brain fog or are taking strong medication, there can be problems with communication and memory and falling asleep during their journey.

Suffers are required to put lots of planning into journey planning due to their physical conditions. Consider any impact of any associated mental health.

### Remember in PIP...

Can they do this activity safely? How do they manage familiar and unfamiliar journeys? How would they cope with a diversion? Can they use public transport? Also, do they have any associated anxiety secondary to their condition? If so, does this meet the threshold of OPD? If it does, can they manage any journeys alone? What symptoms do they experience? How do they overcome these? Could they go out on the majority of days with support?

## Activity 12: Moving Around

Mobility aids and wheelchairs may be used most or all the time. Often limited movement in their legs and slower movements, they require frequent rest breaks due to pain and fatigue. Use of medication to mask pain can lead to overdoing tasks causing inflammation and increased pain limiting mobility further. Overdoing tasks can increase a patients risk of having a stress fracture. Terrain and weather conditions impact on claimants greatly. Cold and wet conditions heighten pain intensity with arthritic bones. Heat can cause inflammation. Some claimants may undergo corrective or strengthening surgery with the insertion of rods, plates and screws or wear splints or braces and often find it to be temporary and sometimes re-occur at an accelerated level. Standing for lengths of time can cause extreme pain, particularly in long supermarket/shop queues. It is very challenging for patients to get to the supermarket to do their shopping as they may be unable to push trolleys. Some patients may require an adapted mobility car to suit their needs.

### Remember in PIP...

Lived examples help to paint a clear picture of function to the Department. Are they wheelchair bound? If they can walk, where can they walk? How long does it take? Do they have falls? Can they repeat the distance? Do they use aids? Do they have pain? How do they feel after? Does their fatigue increase? Do they have frequent fractures?



## Additional reading or other resources

### EXTERNAL

- <https://FDSSUK.org.uk>
- <https://www.fdssuk.org.uk/guidelines>
- <https://www.magicfoundation.org/Growth-Disorders/McCune-Albright-Syndrome-or-Fibrous-Dysplasia/>

### INTERNAL

- Desktop Aid – Fatigue, Activity 12, MSE, Activity 6

## Version control