Condition Insight Report (CIR)

Alpha-1

Version 1.0

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Overview

What is the condition usually called/ any abbreviations used?

- Alpha-1
- Antitrypsin Deficiency
- A1AD
- AATD

Brief overview of the condition

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a genetic (inherited) condition – it is passed from parents to their children through their genes. Alpha-1 may result in serious lung disease in adults and/or liver disease at any age.

For each trait a person inherits, there are usually two genes; one gene comes from each parent. People with Alpha-1 have received two abnormal alpha-1 antitrypsin genes. One of these abnormal genes came from their mother and one from their father.

When AAT levels are reduced or absent, the balance between AAT and the enzyme elastase is thrown off and can cause damage. Normally, this enzyme plays an important role in fighting infection, but too much of it can harm healthy tissue. It causes damage to the lining and alveoli of the lung, resulting in emphysema, or permanent enlargement of the lung's airways. This can cause problems with the exchange of gas and clearance of mucus from the lung. Additionally, AAT deficiency can affect the liver, leading to poor function and increasing the risk of cirrhosis and liver cancer. In the first three decades of life, liver disease is more common than lung disease for a person with AAT deficiency. In some individuals, AAT deficiency may cause frequent red, painful nodules on the skin.

Presenting Symptoms

Symptoms can appear early in life, but many will not begin until a person reaches middle-age.
Individuals with AAT deficiency have a wide variety of symptoms which may include:
•Shortness of breath
•Excessive cough with phlegm/sputum production
•Wheezing
•Decrease in exercise capacity and a persistent low energy state or tiredness
•Chest pain that increases when breathing in

Symptoms may be chronic or occur with acute respiratory tract infections, such as a cold or the flu.

When the liver is affected by AAT deficiency, symptoms may include;

- Tiredness
- Loss of appetite
- Weight loss
- Swelling of the feet or belly
- Yellowish discoloration of the skin (jaundice) or white part of the eyes,
- Vomiting of blood
- Blood in stools.

In rare cases, AAT can cause a skin disease called panniculitis, resulting in hardened patches and red, painful lumps.

1 in every 3,000 to 5,000 people have Alpha-1

Treatments

Patients who are diagnosed with AAT deficiency before symptoms occur usually have better outcomes than those who are diagnosed at later stages.

If a claimant has low to no symptoms, they may be advised to return for regular follow-ups and make lifestyle changes surrounding alcohol consumption, diet and smoking.

If AAT deficiency has caused lung disease, treatment may include;

- Prescribed inhaled medications to control symptoms of COPD,
- Referral to a pulmonary rehabilitation program,
- Oxygen therapy
- Antibiotics and inhaled corticosteroids to control symptoms of flare-ups, infections or exacerbations if needed.

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?
If someone has developed liver cirrhosis as a result of their Alpha-1, this can cause low platelet levels which makes it harder for your blood to clot. This may make chopping and cutting in the kitchen more risk for the claimant.	If a claimant reports they are suffering from breathlessness or fatigue, then it is likely these symptoms may impact ability to do activities in a timely manner. You must explore if they report they can complete activities, how long is it taking them? Is it more than double the time of someone without a disabling condition?	We must remember that people with this condition will often have suffered from symptoms for a prolonged period and may consider themselves to have adapted. Consequently, we must ensure they are doing activities to an acceptable standard. How does doing things like washing, cooking and walking make them feel?	With conditions where fatigue and breathlessness are present, repeatability is extremely important. How do they feel after completing an activity? How long does it take to recover? Could they move onto another activity? Could they repeat as many times as reasonably expected throughout the day?

Did you know?

More often than not, someone is diagnosed with A1AD because they have become ill. In the case of lung disease, this often happens so slowly that the patient doesn't actually get a diagnosis until around half of lung function has been lost.

Watch a video here by the chair of Alpha-1 UK support group

A mis-diagnosis of "adult onset asthma" is very common. The early symptoms of cough, wheeze and a tendency to develop chest infections are difficult to separate from asthma.

Customer Care

- Remember to **give someone time** to respond and ensure they do not feel rushed
- **Do not** make assumptions around symptoms of treatment
- Ensure if they have any difficulties with finishing sentences due to their condition, you. **encourage companion contribution**
- If completing a face to face assessment, ensure there is **enough air circulating** in your assessment room for the claimant to feel comfortable.

REMEMBER: Conditions such as Alpha-1, often go hand in hand with other conditions likely to alter function or lead to other co-morbidities such as cirrhosis of the liver, emphysema and COPD.

Your assessment should be focused on functional ability, not diagnosis.

Functional Impact

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

Activity 1: Preparing food

Someone with Alpha 1 may suffer from fatigue and breathlessness and consequently have difficulties completing this activity.

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?

Activity 2: Taking nutrition

Someone who suffers from Alpha-1 may have a reduced appetite. They may also be receiving supplements for weight loss.

You must also consider the level of their breathlessness or fatigue may impact their ability to reliably take nutrition.

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?

Activity 3: Managing therapy and monitoring a health condition

People with Alpha-1 may be receiving therapy in the home. If they have any chest physiotherapy, they may require assistance with this which must be explored.

They may also be on multiple medications and taking this on a regular basis.

AAT augmentation therapy is also a common treatment, however, this is most likely to be received in a hospital environment.

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?

Functional Impact

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

Activity 4: Washing and Bathing

A person's ability to wash and bathe may be impacted by fatigue and breathlessness secondary to Alpha-1

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?

Activity 5: Managing toileting needs and incontinence

Some of the common medications used to treat Alpha-1 can cause drug induced incontinence.

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

A person's ability to dress and undress reliably may be impacted by their symptoms of fatigue or breathlessness.

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 breathlessness and/or fatigue? Do they require assistance or use any aids? Can they dress reliably whilst seated?

Activity 12 Moving Around

Remember in PIP…

You must explore STAR when asking about someone's ability to mobilise. It is not sufficient to simply indicate a distance or time someone can walk for.

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 breathlessness/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?

Functional Impact

Although Alpha-1 itself may not impact these activities within the scope of the PIPAG, please ensure you explore any co-morbidities that might.

Activity 7: Communicating Verbally

Remember in PIP…

Can someone express and understand both basic and complex verbal information? Who do they speak to? Can they use a mobile phone? Do they have any cognitive or sensory impairments that may impact their ability to complete this activity?

Activity 8: Reading and understanding signs and symbols

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

Remember in PIP…

Who do they engage with on a regular basis? How do they feel meeting unfamiliar people? If they have anxiety with unfamiliar people, who can support them?

Functional Impact

Although Alpha-1 itself may not impact these activities within the scope of the PIPAG, please ensure you explore any co-morbidities that might.

Activity 10: Budgeting

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 there area impacted by low mood or any cognitive impairments?

Activity 11: Planning and following a journey

Remember in PIP…

You must explore someone's ability to complete both familiar and unfamiliar journeys. If someone reports the presence of anxiety, probing questions should be asked around the potential of OPD. This would include what their symptoms are, how they manage them, if they had support could they complete journeys on the majority of days? Furthermore, general questions such as ability to follow a diversion, ability to plan a journey, ability to use public transport. Remember, physical restrictions are not covered within the scope of Activity 11.

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Additional reading or other resources

EXTERNAL

What is Alpha-1 - Alpha-1 Foundation (alpha1.org)

Alpha-1 UK Support Group – Supporting alphas, their friends and families since 1997 (alpha1.org.uk)

Alpha-1 Antitrypsin Deficiency - causes, symptoms, diagnosis, treatment, pathology – YouTube

Alpha-1 Antitrypsin Deficiency | Patient

INTERNAL

Desktop-Aid-Activity-12

Desktop-Aid-Fatigue

VERSION CONTROL