A guide to completing the ASUK CoRDS Patient Registry

**Introduction**

Alström Syndrome UK would like to provide as much help as we can to support participants with the process of joining the Patient Registry.

This guide will provide:

Detailed guidance notes explaining what each question is asking for.

Tips on the type of information it will be useful to prepare before completing the questionnaire.

An explanation of medical terms, in particular those that are not commonly used.

Definitions for the acronyms used within the questionnaire.

More information about the Patient Registry can be found here:

<http://www.alstrom.org.uk/patient-registry/>

Or feel free to contact Catherine Lewis on

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**Top Tips**

If you make any mistakes you will be able to delete them, and the system will automatically save your changes.

The ASUK questions are not mandatory – you can skip those that you would prefer not to answer or those you feel do not apply to you.

To help you complete this questionnaire it may be helpful if you have the following information readily available before you start:

Details of medication or treatment that the participant is currently taking or has taken in the past

The participants GP practice name and address

The participants medical history, medical notes and details of any hospital admissions.

If any of your answers change AFTER you have completed the questionnaire you can log back in at ANY time to update them.

**Guidance Notes**

**Question 1** - We would like to understand how the participant was first diagnosed with Alström Syndrome and what were the first symptoms that led the participant participant’s parents or carers to seek medical attention.

**Question 2** – We would like to know whether the participant was born on their due date. Please select either YES or NO. If you don’t know the answer you can skip to Question 3.

**Question 3** – If the participant was born early please tell us by how many days for example 10 days early. If the participant was not born early or you are unsure you can skip to Question 4.

**Question 4** – If the participant was born late please tell us by how many days for example 10 days late. If the participant was not born late or you are unsure you can skip to Question 5.

**Question 5** – Please tell us the age of the participants Mother when the participant was born.

**Question 6** – Please tell us the participants weight at birth.

**Question 7** - We would like to understand if any health professional had any concerns about the participant in the first 12 months of their life. By health professional we mean someone that provides health care treatment and advice based on formal training and experience. This includes those who work as a GP, Doctor, Nurse, Optician, Midwife, Health Visitor or Pediatrician.

**Question 8** – Please tell us when the participant first started to walk independently.

**Question 9** – Please tell us when the participant was able to walk confidently.

**Question 10** – Please tell us how the participant was fed as a baby. Mixed feeding refers to a combination of breast feeding and bottle feeding.

**Question 11** – We would like to understand when the participant was first introduced to solid food.

**Question 12** – Please provide as much information as you can about the medicine the participant is currently taking and when and why there may have been changes to the dosage.

**Question 13** - We would like to know if the participant has been diagnosed with a heart condition. Please select either yes or no.

**Question 14** – Please provide more information about the participants heart condition either by selecting one of the heart conditions listed or by selecting ‘other’. If you select ‘other’, please add details

**Dilated cardiomyopathy** is where the chambers of the heart are enlarged because the heart muscle is weakened and cannot pump effectively

**Ischemic heart disease** means that the heart is not getting enough blood and oxygen due to narrowed arteries that supply blood to the heart muscle. Ischemic heart disease is also sometimes called coronary heart disease (CHD).

**Pulmonary hypertension** is a type of high blood pressure that makes it harder for blood to flow through the arteries in your lungs and the right side of your heart.

**Restricted cardiomyopathy** is when the walls of the lower chambers of your heart (called the ventricles) are too stiff to expand as they fill with blood. The pumping ability of the ventricles may be normal, but it’s harder for the ventricles to get enough blood.

**Question 15 –** Please tell us when the participant was first diagnosed with a heart condition. If the participant has not been diagnosed with a heart condition you can skip to the question 19.

**Question 16 –** We would like some more information about the symptoms the participant experienced that led to a diagnosis either by selecting one (or more) of the conditions listed or by selecting ‘other’ if the specific condition is not included within the list.

**Shortness of breath** is also described as a tightening in the chest, difficulty breathing, breathlessness or a feeling of suffocation.

**Poor feeding (in infants)** describes a baby with little interest in feeding. It can also refer to a baby who is not feeding enough to receive the necessary nutrition required to grow.

**Poor growth (in infants)** describes a baby or child whose weight, or rate of weight gain, is below that expected of children of a similar age and gender.

**Fatigue** is when tiredness is often overwhelming and isn't relieved by sleep and rest

**Raised blood pressure** is also sometimes called high blood pressure.It is when the force of your blood pushing against the walls of your blood vessels, is consistently too high**.**

**Repeated respiratory tract infections (RTIs)** is when there are repeated infections of the sinuses, throat, airways or lungs. Typical symptoms can include cough, sore throat, runny nose, nasal congestion, headache, fever, facial pressure and sneezing.

**A blue tinge to the skin (cyanosis)** is when the lips, tongue, face or skin suddenly turn blue/grey (or gums and around the eyes in darker skin), it is also usually accompanied by difficulty breathing.

**Swelling in the hands, ankles or feet** is when a part of your body enlarges. It's usually the result of inflammation or a buildup of fluid**.**

**Question 17 –** We would like to understand if the participant has received treatment or surgery to relieve symptoms as a result of their heart condition. Please select either yes or no.

**Question 18 –** If you answered ‘yes’ to question 17 we would like some more information about the treatment or surgery the participant has received. Please select one (or more) of the treatments listed or select other if the specific treatment is not included in the list.

**Heart transplant** is surgery to remove the diseased heart from a person and replace it with a healthy one from an organ donor.

**Pacemaker implantation** is the insertion of a small electronic device that is usually placed in the chest to help regulate problems with the heart.

**Defibrillator implantation** is a small battery-powered device placed in the chest to monitor heart rhythm and detect irregular heartbeats. It can also deliver electric shocks to fix an abnormal heart rhythm.

**Beta-blockers to treat an irregular heartbeat or heart failure –** this medication reduces blood pressure and helps the heart to beat more slowly and with less force.

**Diuretics** are a type of treatment for high blood pressure that help to remove excess fluid from your body.

**Anticoagulants** are a type of medication that helps to prevent blood clots.

**Question 19 –** Please tell us if the participant has a condition that affects their eyesight. Simply select either 'yes' or 'no'. If your answer is no skip to question 23.

**Question 20 –** Please select 1 of the 3 options listed to describe the participants vision.

**Question 21 –** Please tell us when the participants vision loss was first diagnosed.

**Question 22 –** Please select 1 of the 3 options listed that best describes the participants sight.

**Question 23 –** Photophobia is when the eyes are very sensitive to light. It can involve an uncomfortable or painful feeling in response to sunlight or indoor lighting. Please tell us if the participant has been diagnosed with this condition by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ you can skip to question 25.

**Question 24 -** Please tell us when the participant was first diagnosed with Photophobia.

**Question 25 –** Nystagmus is when the eyes make repetitive, uncontrolled movements (wobble). These movements often result in reduced vision and depth perception and can affect balance and coordination. Please tell us if the participant has been diagnosed with this condition by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ you can skip to question 27.

**Question 26 -** Please tell us when the participant was first diagnosed with Nystagmus.

**Question 27 –** Cone Rod Dystrophy describes a group of eye disorders that affect the light sensitive cells of the retina called the cones and rods. First signs and symptoms usually occur in childhood and may include decreased sharpness of vision and sensitivity to light. Please tell us if the participant has been diagnosed with this condition by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ you can skip to question 29.

**Question 28** - Please tell us when the participant was first diagnosed with Cone Rod Dystrophy.

**Question 29** - Please tell us if the participant has hearing loss. Simply select either 'yes' or 'no'. If your answer is no skip to question 32.

**Question 30** - Please select 1 of the 5 options listed that best describes the participants hearing loss.

**Question 31** - Please tell us when the participants hearing loss was first diagnosed.

**Question 32** – Glue ear is a condition where the middle part of the ear fills with sticky like fluid instead of air. This causes muffled hearing. Please tell us if the participant has been diagnosed with this condition by selecting either ‘yes’ or ‘no’.

**Question 33** - Please select 1 of the 3 options listed that best describes the participants hearing loss.

**Question 34** – Please tell us if the participant uses hearing aids and select 1 of 3 options to inform us how often they are used. If hearing aids are not used at all skip to question 36.

**Question 35** – Please tell us when the participant first started using a hearing aid.

**Question 36** – A cochlear implant is an electronic device that helps to restore some hearing. The device bypasses the damaged parts of the ear to deliver sound signals to the auditory (hearing) nerve. Please tell us if the participant has a cochlear implant(s) by selecting either ‘yes’ or ‘no’. If the answer is ‘no’ skip to question 39.

**Question 37** – Please tell us when the participant first had a cochlear implant fitted

**Question 38 –** Select 1 of 3 options to tell us which side the cochlear implant is fitted or if the cochlear implant is fitted on both the left AND right sides.

**Question 39 –**Type 2 Diabetes is a condition in which cells cannot use blood sugar (glucose) efficiently for energy. This happens when the cells become insensitive to insulin and the blood sugar gradually gets too high. Please tell us if the participant has a Diabetes Type 2 diagnosis by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ skip to question 41.

**Question 40 -** Please tell us when the participant was first diagnosed with Type 2 Diabetes.

**Question 41 -** Insulin resistance is when cells of the body don’t respond properly to the hormone insulin and are less able to lower the level of glucose (sugar) in the blood. This causes too much sugar to stay in the blood. Please select either ‘yes’ or ‘no’ to tell us if you have been diagnosed with this condition.

**Question 42 -** Type 1 Diabetes is a condition where your blood glucose (sugar) level is too high because your body can't make a hormone called insulin. Please tell us if the participant has a Diabetes Type 1 diagnosis by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ skip to question 44.

**Question 43 -** Please tell us when the participant was first diagnosed with Type 1 Diabetes.

**Question 44 -** Scoliosis is a condition that causes sideways curve of the spine, or backbone. Please tell us if the participant has been diagnosed with Scoliosis by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ skip to question 48.

**Question 45 -** Please tell us when the participant was first diagnosed with Scoliosis.

**Question 46 -** We would like to understand if the participant has received treatment to relieve symptoms of Scoliosis. Please select either 'yes' or 'no'.

**Question 47 -** We would like to understand if the participant has received surgery to relieve symptoms of Scoliosis. Please select either 'yes' or 'no'.

**Question 48 -** Kyphosis is a spinal disorder in which an excessive outward curve of the spine results in an abnormal rounding of the upper back. Please tell us if the participant has been diagnosed with Kyphosis by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ skip to question 52.

**Question 49 -** Please tell us when the participant was first diagnosed with Kyphosis.

**Question 50 -** We would like to understand if the participant has received treatment to relieve symptoms of Kyphosis. Please select either 'yes' or 'no'.

**Question 51 -** We would like to understand if the participant has received surgery to relieve symptoms of Kyphosis. Please select either 'yes' or 'no'.

**Question 52 -** Non-alcoholic fatty liver disease (NAFLD) is a condition that causes a buildup of excess fat in the liver of people who drink little or no alcohol. Please tell us if the participant has been diagnosed with NAFLD by selecting either 'yes' or 'no'. If your answer is 'no' skip to question 56.

**Question 53 -** Please tell us when the participant was first diagnosed with NAFLD.

**Question 54 -** We would like to understand if the participant has received surgery or treatment to relieve symptoms of NAFLD. Please select either 'yes' or 'no'.

**Question 55 –** If you answered ‘yes’ to question 54 we would like some more information about the treatment or surgery the participant has received. Please select one (or more) of the treatments/surgeries listed or select ‘other’ if the specific treatment/surgery is not included within the list.

**Question 56 –** Please tell us if the participant has ever had any problems with their kidneys by selecting either ‘yes’ or ‘no’. This includes symptoms that have occurred as a result of a poor kidney function such as:

Swollen ankles, feet or hands

Shortness of breath

Tiredness

Blood in your pee (urine)

An increased need to pee (particularly at night)

Difficulty sleeping (insomnia)

Itchy skin

Muscle cramps

Feeling sick

Headaches.

**Question 57 –** Kidney dialysis is a procedure that removes waste products and excess fluid from the blood when the kidneys stop working properly. It often involves transferring blood to a machine to be cleaned. Please tell us if the participant has ever received kidney dialysis by selecting either 'yes' or 'no'.

**Question 58 –** Please tell us how old the participant was when they first started experiencing symptoms as a result of the kidneys not working properly.

**Question 59 –** We would like to understand if the participant has received surgery or treatment to relieve the symptoms associated with poor kidney function. Please select either 'yes' or 'no'.

**Question 60 –** If you answered ‘yes’ to question 59 we would like some more information about the treatment or surgery the participant has received. Please select one (or more) of the treatments/surgeries listed or select ‘other’ if the specific treatment/surgery is not included within the list.

**Question 61 –** A urological problem refers to an issue with either the kidneys, the ureter (the tubes through which urine flows from the kidneys to the bladder), the bladder (the sac that stores urine), the urethra (the tube through which urine travels from the bladder out of the body) or the adrenal glands (that release hormones). We have provided a sample of common urological conditions, please select the one(s) that apply to the participant or select ‘other’ if the urological condition experienced isn’t listed.

**Question 62 -** We would like to understand if the participant has received surgery or treatment to relieve any urological problems experienced. Please select either 'yes' or 'no'. If your answer is ‘no’ you can skip to question 64.

**Question 63 -** If you answered ‘yes’ to question 62 we would like some more information about the treatment or surgery the participant has received. Please select one (or more) of the treatments/surgeries listed or select ‘other’ if the specific treatment/surgery is not included within the list.

**Question 64 –** *Questions 64 and 65 apply to female participants only. Male participants can skip to question 66.* A gynecological problem refers to any issue with female reproductive organs. Please indicate if the participant experiences any gynecological problems by selecting either ‘yes’ or ‘no’. If your answer is no you can skip to question 66.

**Question 65 -** We would like to understand if the participant has received surgery or treatment to relieve any gynecological problems experienced. Please select either 'yes' or 'no'. If your answer is ‘yes’ please tell us about the treatment or surgery the participant has received either by selecting one (or more) of the treatments/surgeries listed or by selecting ‘other’ if the specific treatment/surgery is not included within the list.

**Question 66 -** Delayed puberty is a term that explains a lack of testicular enlargement in boys and a lack of breasts and menstrual periods in girls. Please tell us if the participant has experienced a delay to puberty by selecting either 'yes' or 'no'. If your answer is 'no' skip to question 68.

**Question 67 -** We would like to understand if the participant has received treatment to address a delay to puberty. Please select either 'yes' or 'no'.

**Question 68 –** A learning difficulty can affect the way a person understands information and how they communicate. This means they can have difficulty understanding new or complex information, learning new skills or coping independently. Please indicate if the participant has been diagnosed with a learning difficulty by selecting ‘yes’ or no’. If your answer is ‘no’ you can skip to question 70.

**Question 69 -** If you answered ‘yes’ to question 68 we would like some more information about the learning difficulty the participant has been diagnosed with. Please select one (or more) of the conditions listed or select ‘other’ if the specific learning difficulty experienced is not included within the list.

**Question 70 –** There are many known conditions that typically occur in individuals diagnosed with Alström Syndrome. We would like to understand which of these the participant is affected by. Using the list please select the conditions that are relevant to the participant. If there are any conditions that the participant is affected by that are not provided within the list please then select ‘other’.

**Question 71 –** An organ transplant is surgery to remove a diseased organ from a person and replace it with a healthy one from an organ donor. Please tell if the participant has received an organ transplant by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ skip to question 74.

**Question 72 –** Using the list provided please indicate which organ transplant(s) the participant has had.

**Question 73 –** Please tell us the age of the participant when they had surgery(ies) to transplant an organ (s).

**Question 74 –** Please tell us if the participant has ever been admitted to hospital (including admissions not related to Alström Syndrome), you can do this by selecting either ‘yes’ or ‘no’. If your answer is ‘no’ you can skip to question 77.

**Question 75 –** Please tell us how many times the participant has been admitted to hospital.

**Question 76 –** Please tell us about why the participant was admitted to hospital, you can select more than one reason. If the reason is not included in the list, then please select ‘other’.

**Question 77 –** Please tell us the participant’s current weight. We have asked for weight to be provided in kilograms but if you are not familiar with kilograms there is a tool available online that can convert your weight into a format that is suitable for the registry. Click here for help: <https://www.thecalculatorsite.com/conversions/common/kg-to-stones-pounds.php>

**Question 78 -** Please tell us the participant’s current height. We have asked for height to be provided in centimetres but if you are not familiar with using centimetres there is a tool available online that can convert your height into centimetres. Click here for help: <https://www.thecalculatorsite.com/conversions/common/cm-to-feet-inches.php>

**Question 79 –** Please tell us about how often you exercise. When we use the term exercise we mean any physical activity that involves movement of the body and using energy. Some examples of exercising include walking, gardening, riding a bike, climbing the stairs, playing sports or dancing - these are all good examples of being active.

**Question 80 –** Please tell us how many portions of fruit and vegetables you eat in a day. As a rough guide, 1 portion is the amount of fruit or vegetables that can fit in the palm of your hand.

**Question 81 –** Please tell us how often the participant eats sweets, chocolate and crisps or any food that is very high in salt, fat or sugar.

**Question 82 -** Please tell us how often the participant has drinks that are very high in sugar such as Coke, Sprite, Fanta etc.

**Question 83 –** A person with a healthy diet will eat at least 5 portions of a variety of fruit and vegetables every day and consume a variety of foods that provide the necessary nutrients needed to maintain health and have energy. These nutrients include protein, carbohydrates, fat, water, vitamins, and minerals. A healthy diet also means only having very small amounts of food and drink that are high in fat, salt and sugar. Using this information as your guide please tell us about the diet of the participant and the extent to which it is ‘healthy’.

**Question 84 –** Please tell us about your ethnicity as this helps us to understand the diversity of patients and provide better and more appropriate services.

**Question 85 –** If there is any other information you feel has not been captured by this questionnaire and you would like to tell us about it then please do so here.