Alström Syndrome

A guide to greater understanding of this rare condition

For consultants, GPs and other health professionals as well as parents seeking to know more about Alström Syndrome
Alström Syndrome UK, a registered charity, was established in 1998 to provide support for families, professionals and carers.

The charity has been instrumental in developing specialised medical screening clinics in both Birmingham and in Torquay for those affected. Here families get advice on the best treatments available to maintain a good quality of life and delay progression on the disorder.

Each year a number of combined multi-disciplinary clinics are organised - one alongside the annual family Alström Syndrome UK conference.

The conference gives families a chance to hear talks from specialists on how best to manage the disorder and meet others affected.

For the latest information visit: www.alstrom.org.uk

Alström Syndrome UK founder & Chief Executive Kay Parkinson

This booklet has been produced by Alström Syndrome UK in an effort to promote greater awareness and understanding of Alström Syndrome, and to widen the spread of current knowledge. It has been funded by the NHS National Specialised Commissioning Group (NCG).

It is hoped that the booklet will prove useful to medical and healthcare professionals and to parents anxious to know more about the Syndrome and the advances in methods of treatment.

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This booklet uses a wide range of terms for medical conditions which may affect children and adults with Alström Syndrome. These are explained throughout the booklet and summarised in a glossary on page 17. Please note that the symptoms and conditions listed are intended to aid diagnoses and do not necessarily apply in every case of Alström Syndrome.
Introduction to Alström

Alström Syndrome is a very rare recessively inherited condition and the symptoms are not always easily recognised.

Because of its rarity, many GPs, doctors working in hospitals and many other health professionals will not have come across Alström before - or perhaps have even heard of it.

It is estimated that about 200 families in the UK could be suffering from Alström Syndrome - but many will not have been identified as having the symptoms and so will be missing out on the significant advances being made in this area.

This booklet is designed to create greater awareness and understanding of the Alström Syndrome throughout the Health Service, to provide guidance and to highlight the help that is currently available to both children and adults to enable them to lead longer and more fulfilled lives.

What is Alström?

Alström Syndrome is characterised principally by a number of key conditions:

- Retinal degeneration (inherited progressive eye disease)
- Sensorineural hearing loss (disorders of the cochlear part of the ear)
- Obesity and Insulin resistance

Additional features can include:

- Renal and hepatic dysfunction (affecting the kidneys and liver)
- Type 2 diabetes mellitus
- Hypertriglyceridaemia (elevation of fatty substances found in the bloodstream)
- Cardiomyopathy (poor cardiac function where the heart muscle is weakened and enlarged)
- Bladder and bowel problems

The origins of Alström Syndrome

Alström Syndrome has similar characteristics to the Laurence-Moon-Bardet-Biedl Syndrome. Important differences between the two were first discovered by Carl-Henry Alström and three associates in 1946; further detailed investigations on three patients formed the basis of a very thorough manuscript in 1959.

It detailed the apparently recessive hereditary combination of retinal degeneration, obesity, sensorineural hearing loss and diabetes found in Alström.

“I am sure Carl-Henry Alström would be delighted to learn of the work of Alström Syndrome UK and the worldwide research currently being undertaken,” says ASUK founder and Chief Executive Kay Parkinson.
What causes Alström?

The condition is recessive, with mutations in both copies of the gene ALMS1. Each parent will have one normal copy of the gene and one with a significant mutation (carrier status).

On average one in four children of a couple who are both carriers will inherit both ALMS1 genes with mistakes and suffer from the condition. More than 80 mutations in the ALMS1 gene have been identified in families with Alström syndrome.

The protein ALMS1, whose function is currently unknown, localises to the centrosome and/or the ciliary basal body. This localisation has implicated Alström syndrome as one of a growing collection of disorders known as ‘ciliopathies’.

Cilia are ancient, evolutionarily conserved organelles that project from cell surfaces to perform diverse biological roles. Defects in cilia are associated with a range of human diseases, such as primary ciliary dyskinesia, hydrocephalus, polycystic liver and kidney disease, and some forms of retinal degeneration. Recent evidence indicates that ciliary defects can lead to a broader set of developmental and adult phenotypes, with mutations in ciliary proteins now associated with nephronophthisis, Bardet-Biedl Syndrome, Alström Syndrome, and Meckel-Gruber Syndrome.

Early diagnosis of these ‘ciliopathies’ allows more appropriate clinical intervention, which may enable improvements in quality of life and longevity.

Medical terms explained

- Centrosome (tiny organising centre of the cell)
- Ciliopathies (a genetic disorder of the cellular cilia - cilia are essential to many of the body’s organs)
- Organelles (components of cells)
- Ciliary dyskinesia (progressive disorder affecting organ systems)
- Hydrocephalus (‘water on the brain’)
- Adult phenotypes (characteristics displayed by an organism)
- Nephronophthisis and nephropathy (structures and function of the kidneys)

Alström Syndrome UK charity and two centres have been designated by the National Commissioning Group, an NHS service for highly-specialised services, to provide experts in Alström Syndrome and multi-disciplinary clinics for Alström patients.

Alström Syndrome UK - Family Support Services and National Co-ordination of clinics:

- Birmingham Children’s Hospital clinics for under-16s
- Torbay Hospital, Devon, clinics for over-16s

To find out more about the clinics (see also pages 14 & 15):

Email: info@alstrom.org.uk / Tel: 01803 524238 or visit the Alström website www.alstrom.org.uk
Good medical practice involves a clear history, physical examination and investigation in any outpatient review (see our website for more comprehensive guidance).

**History from Alström person and family**
- General well-being
- Psychology - adjustment to condition
- Mobility
- Eyesight and visual aids - Braille, IT
- Hearing and appropriate hearing aids
- Growth and development

**Ask if there is any:**
- Shortness of breath
- Tiredness
- Problems with passing water
- Heartburn
- Treatment being taken

**Examination**
- Observe mobility
- Weight, height, blood pressure
- Acanthosis (darkening and increase in thickness of cells in the skin)
- Condition of feet
- Listen to heart and lungs
- Diet and exercise assessment

**Blood tests**
- Urea and electrolytes
- Creatinine - full blood count
- Liver function tests
- Haemoglobin A1c and blood glucose
- Serum triglycerides and cholesterol
- Thyroid function
- Oestrogen or testosterone, FSH and LH
- Brain natriuretic peptide

**Scans and imaging**
- Bladder scan post micturition (after the passage of urine) if urinary symptoms
- ECG
- Echocardiogram
- Respiratory function
- Liver scan

**From seven years and older standard respiratory function tests**
- Peak Flow
- Forced Expiratory Volume in 1 second (FEV)
- Forced Vital Capacity (FVC)
- FEV1/FVC ratio
- Digital oximetry (determination of oxygen and protein levels in the blood) at rest and after a five-minute walk
The following key factors distinguish Alström Syndrome from most other cone rod dystrophies:

- Nystagmus and photophobia, usually but not always, in early infancy.
- Progressive pigmentary retinopathy (cone-rod dystrophy) leading to blindness.
- Childhood obesity, often moderating to high-normal weight in adulthood.
- Mild to moderate bilateral sensorineural hearing loss.
- Congestive heart failure secondary to cardiomyopathy, in infancy or early adulthood.
- Hyperinsulinemia / insulin resistance.
- Non-insulin dependent diabetes mellitus (Type 2 diabetes or NIDDM) developing in early adulthood.
- Progressive chronic nephropathy.
- Normal intelligence with some reports of delayed early developmental milestones.

A wide range of other conditions can be present in some, but not all, cases including hypothyroidism, portal hypertension, alopecia, scoliosis, hypertension, urinary tract infections, gastro-intestinal reflux, asthma and respiratory problems (see the website www.alstrom.org.uk for a more comprehensive guide).

As the basic biochemical aetiology of Alström is unknown, there is currently, no cure. It is more realistic to concentrate approaches to treatments on specific organ systems.

Symptoms of Alström Syndrome vary considerably, even among siblings.

Medical terms explained

- Nystagmus [wobbly eyes]
- Photophobia [dislike of light]
- Cone-Rod Dystrophy (CRD) [an inherited progressive eye disease]
- Cardiomyopathy [where there are defective heart cells]
- Nephropathy [kidney disease]
- Aetiology [study of causes and origins]
Eyes

The first feature often noticed are the eyes. Affected children have an involuntary rapid movement of the eye (wobbly eyes / nystagmus) and extreme photophobia (extreme sensitivity to light).

Poor vision from infancy can also be present and eventually lead to a degeneration of the retina, the thin, light sensitive lining at the back of the eye (retinopathy) and blindness.

Alström Syndrome is one of a number of causes of cone-rod dystrophy (CRD). Protection of the retina from bright light with dark glasses may slow down visual loss.

Symptoms of CRD are seen as decreased visual acuity in the early stages followed by loss of peripheral vision.

Patterns of visual loss vary in Alström Syndrome and other CRD such as retinitis pigmentation.

Treatment

A cure may eventually come from advances in gene replacement therapy. In the meantime, people with CRD can find comfort by protecting their retinas from bright light.

Currently, there is nothing proven to prevent visual deterioration. Ophthalmological advice is crucial to optimise life with low vision. Dark glasses will be needed indoors and out to combat the extreme photophobia experienced by Alström patients when young. Prescription glasses can also help.

Hearing Loss

Sensorineural hearing loss usually begins before the children reach the age of 10. Most sensory hearing loss is due to poor hair cell function. The hair cells may be abnormal at birth, or damaged during the lifetime of an individual. There are both external causes of damage - like noise trauma and infection - and intrinsic abnormalities, like deafness genes.

Hearing loss in Alström patients varies considerably in degree of severity and also age of onset. ‘Glue ear’ is common in childhood, often accompanied by very runny noses.

Treatment

Early recognition is vital to maintain educational and social development and regular hearing tests are advised. Hearing aids and other auditory devices can help considerably in alleviating problems and the patients should be fitted with the latest available models.

Medical terms explained

Nystagmus [wobbly eyes] | Photophobia [dislike of light]

Sensorineural hearing loss [disorders of the cochlear part of the ear]

Glue ear [The middle ear is connected to the back of the nose by a narrow channel called the Eustachian tube. Any fluid that builds up in the middle ear can be drained away by this tube. If the Eustachian tube becomes blocked or swollen, the vacuum of air that builds up can draw fluid into the middle ear cavity from the mucus that lines the rest of the ear. At first the fluid is thin and watery, but later it can become thick and glue-like].
Alström Syndrome can cause the heart muscle to pump less efficiently, known as cardiomyopathy. This occurs in about 40% of infants with Alström and often recovers, although not completely, and can re-occur in later life.

The heart muscle develops lots of small scars (fibrosis), myocardial fibrosis, usually in a patchy manner. In infants. The heart may stretch or dilate, but in most patients the fibrosis tissue makes the heart stiff and inflexible, termed ‘restrictive’ cardiomyopathy.

Other causes of cardiomyopathy, such as viruses, may be suspected until other signs of Alström develop e.g. wobbly eyes (nystagmus) and photophobia (dislike of light).

Cardiomyopathy can also occur during adolescence when the outcome may not be so good. It is often misdiagnosed as asthma in children and young people because it can cause breathlessness and wheeze.

It is important to recognise Alström’s cardiomyopathy early, so treatment can be started promptly. Alström Syndrome UK charity recommend that patients who have had cardiomyopathy should have:

- An annual cardiac review, with an echocardiogram (echo) - a painless ultrasound scan which looks at the structure and function of the heart.
- An ECG (electrocardiogram) which traces electrical activity in the heart.

Ideally, blood oxygen levels should also be monitored, together with exercise capacity. The role of blood hormone tests and cardiac MRI scanning is being evaluated.

Alström Syndrome UK charity also recommends that even if patients have not had cardiomyopathy they should have annual cardiac review with a detailed discussion of exercise capacity and an ECG where there is any hint of:

- Breathlessness
- Excess fatigue
- Development of asthma

Change in ECG results should trigger an echo. It is helpful if the person doing the echocardiogram knows to look for evidence of ‘restrictive’ cardiomyopathy because the signs are often subtle. New echo techniques (Tissue Doppler) will improve this.

**Treatment**

Alström’s cardiomyopathy affects both the right and left sides of the heart, which is important as treatment may differ depending on which side is more severely affected.

Effective treatments to improve symptoms have included: Digoxin, furosemide, angiotensin converting enzyme (ACE) inhibitors, beta-blockers and spironolactone.

Both patients and their doctors should be aware that Alström’s cardiomyopathy can happen suddenly and patients are vulnerable during intercurrent illness and surgery. At these times, close monitoring is essential and expert advice should be sought as needed.
Obesity

Alström children gain weight rapidly, though they may not eat more than their peers; however, they may have limited ability for physical exercise due to decreasing eyesight. Help from a dietitian should be sought at an early age for specific advice.

Adults have been seen to benefit from low carbohydrate diets. Obesity tends to diminish with age.

**Treatment**

Diet and exercise are very important in helping to keep Alström patients weight under control. For adults, low-carbohydrate diets prove effective accompanied by regular exercise. Children may also benefit from a modified healthy eating plan with a reduction in carbohydrate and life long regular exercise is emphasised.

Later, in young adulthood, children become insulin resistant, develop high blood levels of insulin and often type 2 diabetes mellitus in the second or third decade of life.

The diagnosis is often made by blood testing before symptoms of thirst, passing too much urine and weight loss are evident.

**Lipid disorders**

As is common in insulin resistant states Alström subjects tend to have high serum triglyceride levels, low HDL cholesterol and variable increases in total serum cholesterol.

Hypertriglyceridaemia if severe (≥10mmol/l) will respond acutely to fasting and long term to NIASPAN. With milder dyslipidaemia long term statin therapy is indicated to reduce the as yet uncertain risk of atherosclerosis.
Renal failure or kidney failure can occur in Alström. There are acute and chronic forms; either may be due to a large number of other medical problems (including diabetes).

In Alström patients two cases of acute renal failure occurred in patients with ileal conduits (part of the intestine used as a urinary conduit) and chronic kidney failure is more prevalent from the third decade of life. Currently the cause is not fully understood but, like the heart, the kidneys may develop fibrosis.

Renal failure is described as a decrease in the glomerular filtration rate. Each kidney contains over one million tiny filtering units, called glomeruli, which remove waste chemicals etc from the blood.

Problems frequently encountered are abnormal fluid levels in the body, imbalanced pH levels, abnormal levels of potassium, calcium, phosphate, hematuria (blood in the urine) and (in the longer term) anaemia. Long-term kidney problems can impact on other diseases e.g. cardiovascular disease.

Alström Syndrome UK recommends regular blood pressure monitoring, and annual urine testing and serum creatinine testing of all patients (including urea levels).

Acute renal failure

Acute renal failure (ARF) is, as the name implies, a rapidly progressive loss of renal function, generally characterised by oliguria (decreased urine production, quantified as less than 400 ml per day in adults, less than 0.5 ml/kg/h in children or less than 1 m/kg/h in infants); body water and body fluids disturbances; and electrolyte derangement. An underlying cause must be identified to arrest the progress, and dialysis may be necessary to bridge the time gap required for treating these fundamental causes. ARF can result from a large number of causes.

Chronic kidney disease

Stage 5 Chronic Kidney Disease (CKD) can develop slowly and show few initial symptoms, be the long-term result of irreversible acute disease or be part of a disease progression.

Methods of measurement for CKD

Chronic kidney failure is measured in five stages, which are calculated using a patient’s GFR, or glomerular filtration rate. Stage 1 and 2 CKD may be associated with normal kidney function, but increased loss of protein in urine and high blood pressure. However, if there are other signs of damage to the kidneys close monitoring may be necessary. Stages 3 and 4 need increasing levels of supportive care from their medical providers to slow and treat their renal dysfunction.

Patients in stages 4 and 5 usually involve active treatment. Stage 5 CKD is considered a severe illness and requires some form of renal replacement therapy (dialysis) or kidney transplant whenever feasible.

Glomerular filtration rate

A normal GFR varies according to many factors, including sex, age, body size and ethnicity. Renal professionals consider the GFR to be the best overall index of kidney function. The National Kidney Foundation offers an easy to use on-line GFR calculator for anyone who is interested in knowing their glomerular filtration rate (a serum creatinine level, a simple blood test, is needed to use the calculator).

Further information about what these results mean can be found at www.kidney.org.uk.
Transplants

A successful kidney and pancreas transplant has been undertaken on an Alström patient and a number of patients are on waiting lists. Heart transplants to date have not been as successful but this may have been due to a lack of forward planning and the procedure undertaken as an emergency.

Transplants need to be carefully thought through with the expert multi-disciplinary teams at Birmingham and Torbay Hospitals so that all the conditions Alström patients have can be monitored closely.

Endocrine / Fertility / Genitalia

Hypogonadism (defect of the reproductive system) is common amongst affected males with a number having undescended testicles, so tests should be made for hypogonadism. Males may have low testosterone levels and may need medicinal supplements. Females should be tested for polycystic ovaries and a number have needed cysts removed. Thyroid under activity is frequent and easily treated with thyroxine treatment.

Dark patches of skin

Other findings observed in some Alström Syndrome patients include a darkening of areas of the skin, called acanthosis nigricans, almost as though the person has not washed properly. These patches are frequently found on the neck and creases of the elbows.

Other problems where checks are important:

- Pulmonary problems - some patients have smaller than average lungs and can be pre-disposed to bronchial problems

Scoliosis, or curvature of the spine, has been identified in some children, as well as short stature (see next page).

Treatment

Physiotherapy advice should be sought and appropriate exercise undertaken.
Growth and Postural changes

The majority of those with Alström Syndrome have advanced bone age and epiphyseal fusion (maturation of the growing ends of bones at wrist, knee and in the spine) resulting in short stature.

Excessive curvature of the cervical spine (neck) can result in an arching forward of the head and neck. This is reversible in younger subjects but can become fixed in adulthood. This is not seen in other causes of early blindness and affects less than 50%. It is not yet clear whether exercises can prevent this effect.

Developmental spinal changes

A varying degree of forward curvature of the thoracic spine (around the chest area) is extremely common in Alström Syndrome persons. This is rarely severe but may contribute to neck aches and can slightly reduce lung function by restricting lung volume and chest expansion.

In a smaller number, the spine can be twisted sideways (scoliosis). This can also cause pain in the back and if the lower ribs rub against the top of the hip on one side. Regular stretching exercises can relieve discomfort though if kyphoscoliosis (curvature and twisting of the spine) is severe from a young age then major surgery might well be needed.

Secondary spinal changes

Excessive thickening of the spine (spondylitis), which is known to occur sporadically in Type 2 diabetic persons, can rarely occur in Alström Syndrome. If the bony thickening rubs on nerves flowing from the neck, then pains and weakness can exceptionally occur. Surgery for this is very specialised and its place in the syndrome is uncertain.

Arthritis

A minority of young adults have painful joints not yet well characterised.

Therapy

Stretching exercises, swimming, dancing and walking as well as massage - all have a place in relieving symptoms. Weight reduction in obesity cases has also helped to relieve backache.
Blindness due to retinal degeneration and sensorineural partial hearing loss are virtually always accompanied by obesity and insulin resistance. Other manifestations such as infant and/or adult cardiomyopathy, kyphosis and pulmonary fibrosis are variable in occurrence and severity.

This combination of cardio-respiratory changes can predispose to unexpectedly severe hypoxia during episodes of infection or post-operatively. Metabolic disturbances can include hypertriglyceridaemia, and insulin-resistant diabetes.

The severity and responsiveness to treatment of hyperglycaemia is heterogeneous, just as in adult type 2 diabetes as such. A few patients require u 500 insulin in very high doses long term, but many respond to Metformin and/or Glitazones providing cardiac and renal function are good. Hypertriglyceridaemia if severe (→10mmol/l) will respond acutely to fasting and long term to NIASPAN.

In preoperative assessment, therefore, we would advise:

- Echocardiogram
- Pulse oximetry before and after exercise
- Blood glucose
- Serum triglycerides
- Renal and hepatic function tests

Peri-operatively and post-operatively:

Very careful monitoring of heart and oxygenation until fully recovered and ambulant. Monitoring of blood glucose - levels of both blood glucose and serum triglycerides normalise in 24-48 hours during fasting and do not usually require intravenous insulin. NB Photophobia and nystagmus can be troublesome for Alström persons in bright lighting. It is best to plan for a period of ventilation of lungs on intensive care after surgery in Alström persons.

Even for relatively minor procedures under general anaesthetic HDU or ITU should be planned post-operatively.

Pulmonary Oedema:

Alström patients may be more prone to pulmonary oedema, particularly if cardiomyopathy is present. This should be anticipated and planned for and steps such as procedures being undertaken when a dialysis session, for example, is booked for the next day as opposed to having a procedure on a Friday with a weekend in between sessions.

Medical terms explained

Hypertriglyceridaemia and tryglycerides (elevation of fatty substances found in the bloodstream)

Heterogeneous (consisting of elements that are not of the same kind or nature)

Pulmonary oedema (fluid on the lungs)
Alström Syndrome is recessively inherited where both parents are phenotypically normal. Parents who already have an affected child run a risk of further affected children.

Amongst cultures and ethnic groups where cousin marriages are traditional we have found large family groups with affected children. Genetic counselling will help families make the choices that they need to make for their family.

The gene responsible for Alström Syndrome was located by scientists at the Jackson Laboratory in Maine, USA, and scientists at Southampton University, England, simultaneously.

The ALMS1 gene provides instructions for making a protein whose function is unknown. Researchers believe that the protein may play a role in hearing, vision, regulation of body weight, and functions of the heart, kidney, lungs, and liver. It may also affect how the pancreas regulates insulin, a hormone that helps control blood sugar levels (see www.alstrom.org.uk for more guidance).

One fifth of the world’s population live in communities that have consanguineous marriages (of the same blood origin). In the UK, these include those of Pakistani, Middle Eastern, Bangladeshi, Turkish and some groups of Indian origin, plus Irish travellers and some refugees.

In populations where there are consanguineous marriages, recessively inherited disorders - like Alström Syndrome - tend to cluster in extended families that are connected by familial links based on blood ties, consolidated through marriage.

The birth of an affected child is a signal that others in the extended family could have a child with the same disorder. This social context may be ideal for delivering particularly effective genetic testing and counselling services using existing family networks.

An introductory booklet on consanguineous marriage is now available from: Dr Aamra Darr, School of Health, University of Bradford, Unity Building, 25 Trinity Road, Bradford, BD5 0BB.

Tel: 01274 236120 / Email: a.r.darr@bradford.ac.uk
www.brad.ac.uk/communitygenetics

The Genetic Alliance UK can help families who may be worried that they will pass on a genetic condition to their child.

They are able to go to an IVF clinic to conceive their baby in the lab. After a few days, when the embryo has grown to between six and eight cells, one cell is tested for the genetic condition.

If the embryo is free from the condition, it can be implanted in its mother and continue to develop naturally and healthily. The whole process is know as Pre-Implantation Genetic Diagnosis or PGD.

For more details, contact: Genetic Alliance UK, Unit 4D, Leroy House, 436 Essex Road, London, N1 3QP

Tel: 020 7704 3141   Web: www.geneticalliance.org.uk
ASUK has a wealth of information about the Alström Syndrome and keeps up to date with the latest research, as well as providing family liaison support for families who have children affected themselves. They have direct contact with medical experts who will offer advice.

Patients should be encouraged to attend expert patient programmes where they will develop skills to manage their condition.

Tel. 01803 524238 or email info@alstrom.org.uk

The support group was founded by Kay Parkinson in 1998, a working mother from Paignton, Devon, after her two children were diagnosed with Alström Syndrome when aged 18 and 15. Then there was very little known about the condition, no help or support and no specialised medical services.

Today there is a dedicated website giving up-to-date information www.alstrom.org.uk, quarterly newsletters, help and support services, an annual family conference and regular multi-disciplinary clinics at Birmingham Children’s Hospital for children under-16 with the condition and Torbay Hospital for adults.

Alström Syndrome UK charity is an equal partner in providing the Alström clinical services and maintains the only UK database of those affected by this disorder. Our dedicated family liaison officers, themselves parents of affected children provide help and support to families and specific services to Asian families.

Thanks to the fundraising efforts of a dedicated fundraiser, we have now been able to help purchase special equipment and help with the cost of respite holidays for families.

With a £350,000 grant from the Big Lottery, ASUK is now co-ordinating an exciting major research programme involving Cambridge University, Birmingham Children’s Hospital and Torbay Hospital - which offer new hope for those with Alström Syndrome.

Alström Syndrome UK has three key aims:

- To help people with Alström Syndrome - to provide support for them, their carers and the professionals who are working with them.
- To raise awareness amongst both the public and medical professions of Alström Syndrome.
- To raise funds to promote research into Alström Syndrome.

Major research work is being funded by a Big Lottery medical and scientific grant awarded to Alström Syndrome UK

Families getting together at the Alström Family Conference in Torbay
Alström Syndrome UK Support Group (ASUK) was responsible for setting up the world’s first multi-disciplinary clinics for the dedicated care of Alström Syndrome patients. They are based at Birmingham Children’s Hospital and at Torbay Hospital in Devon and have now become an established part of the NHS since adoption by the National Commissioning Group (NCG).

The clinics mean that a wide range of tests and treatments can be carried out under ‘one roof,’ for example:

- A carousel of tests and assessments timetabled for each person with an hour for each of cardiology, respiratory tests, diabetes and metabolism and prearranged appointments for audiology/psychology/urology.

- A summary of results and recommended therapy being sent to each family, and their hospital and community carers.

- A carefully planned set of tests on one blood sample at least once per year (providing patient and family not too upset or young).

Extra visits for specific problems can also be arranged. Accommodation costs will be provided for by ASUK.

If you would like to attend the Birmingham or Torbay clinics, please contact Kay Parkinson of ASUK on 01803 524328 to make arrangements. We hope that families will be helped by attending one or two clinics per year.
Alström Syndrome multi-disciplinary clinics are held at Birmingham Children’s Hospital NHS Foundation Trust, Steelhouse Lane, Birmingham B4 6NH.

Under the clinical lead of Professor Timothy Barrett dedicated multi-disciplinary Alström Clinics are held quarterly at Birmingham Children’s Hospital. Children under 16 can be referred by their GP, Consultant or through Alström Syndrome UK charity for a wide range of clinical investigations known to affect children with Alström Syndrome.

All tests are conducted by the child friendly team who have gained wide experience in treating children affected by Alström Syndrome on the same day saving children and their parents endless trips to hospital.

If you would like to speak to a medical expert in Alström Syndrome here please contact: Professor Tim Barrett, Birmingham Children’s Hospital. Tel: 0121 333 9999.

Torbay Hospital (run by South Devon Healthcare Foundation Trust) in Torquay, Devon, has a dedicated clinic for Alström patients aged 16 and over.

Under the clinical lead of Dr. Richard Paisey, multi-disciplinary clinics are held usually on a quarterly basis, offering Alström patients the opportunity to access a wide range of tests and treatments by a special team in a more relaxed atmosphere. This ‘same day’ service saves an enormous amount of time, travel and stress.

Patients can be referred by their GP, consultant or through the Alström Syndrome UK charity.

If you would like to speak to a medical expert in Alström Syndrome here please contact: Dr Richard Paisey, Torbay Hospital. Tel: 01803 614567.


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**Glossary of medical terms**

- **Acanthosis** (darkening and increase in thickness of cells in the skin)
- **Adult phenotypes** (characteristics displayed by an organism)
- **Aetiology** (study of causes and origins)
- **Centrosome** (tiny organising centre of the cell)
- **Ciliary dyskinesia** (progressive disorder affecting organ systems)
- **Digital oximetry** (determination of oxygen and protein levels in the blood)
- **Dilated cardiomyopathy** (poor cardiac function where the heart muscle is weakened and enlarged)
- **Glomerular filtration** (each kidney contains over one million filtering units called glomeruli which remove waste chemical etc from the blood)
- **Heterogeneous** (consisting of elements that are not of the same kind or nature)
- **Hydrocephalus** (‘water on the brain’)
- **Hypertriglyceridaemia and tryglycerides** (elevation of fatty substances found in the bloodstream)
- **Hypogonadism** (defect of the reproductive system)
- **Ileal conduits** (part of the intestine used as a urinary conduit)
- **Nephronophthisis and nephropathy** (structures and function of the kidneys)
- **Organelles** (components of cells)
- **Post micturition** (after the passage of urine)
- **Pulmonary oedema** (fluid on the lungs)
- **Renal and hepatic dysfunction** (affecting the kidneys and liver)
- **Sensorineural hearing loss** (disorders of the cochlear part of the ear)
An acknowledgement and thanks
by Kay Parkinson

The work of Alström Syndrome UK (ASUK) is greatly supported by Dr Cathy Carey who learnt rapidly about the condition when the late Matthew Parkinson was diagnosed whilst under her care 1998. Dr Richard Paisey was instrumental in developing with ASUK the first multi-disciplinary clinics for Alström Syndrome and greatly supported by staff at Torbay hospital. Professor Tim Barrett at Birmingham Children’s Hospital developed children’s clinical services and is supported by an excellent team.

During 2008, the National Commissioning Group commissioned the partnership services of Alström Syndrome UK, Torbay Hospital and Birmingham Children’s Hospital as a dedicated specialist NHS service.

Thanks also to the wonderful families who despite all the many problems they have to cope with still support the charity with attendance at the clinics and the conference and who have supplied the special photographs in this booklet.

Thanks to my husband John for putting up with me forever in the office or on the phone!

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This booklet is dedicated to the memory of Matthew James Parkinson (1978-2003) and Charlotte Elizabeth Parkinson (1981-2010) and all those diagnosed with Alström Syndrome

“Forever inspired by you”