



**Down's Syndrome
Association**

A Registered Charity No. 1061474

Health Series



Inflammatory Arthritis in Children

A Down's Syndrome Association publication

Our resources and Information Team are here to help

Please see our website for up-to-date information: www.downs-syndrome.org.uk

If you would like to talk about any of the issues raised in this resource, then please get in touch with our helpline by calling 0333 1212 300 or by emailing us on

info@downs-syndrome.org.uk.

Helpline Monday - Friday 10am-4pm | Telephone: 0333 1212 300

The Down's Syndrome Medical Interest Group (DSMIG)

This resource has been produced in collaboration with the Down's Syndrome Medical Interest Group (DSMIG).

DSMIG was launched in 1996 and is a registered charity. It is a network of healthcare professionals – mainly doctors – from the UK and Republic of Ireland whose aim is to share and disseminate information about the medical aspects of Down syndrome and to promote interest in the specialist management of the syndrome.

Arthritis (inflammation of the joints) is an autoimmune condition.

Children with Down's syndrome have an increased risk of autoimmune conditions. Autoimmune conditions are caused by a malfunction in the body's natural defence systems against infections

The small but slowly growing body of evidence suggests there is an increased risk of an inflammatory form of arthritis in children with Down's syndrome.

Health professionals call this condition '*Inflammatory arthritis of Down's syndrome*' (our preferred term). However, you may hear some health professionals refer to this condition as '*Down's arthropathy*'.

Inflammatory arthritis of Down's syndrome is an under-recognised condition that, if untreated, results in chronic disability and functional impairment

Greater awareness of the increased risk of this condition will lead to earlier diagnosis and treatment.

Background Information

A number of autoimmune disorders such as diabetes mellitus, coeliac disease and thyroid dysfunction (typical under active) are well described in people with Down's syndrome. Arthritis (inflammation of the joints) also occurs but is largely under-reported in this population group.

The 'Arthropathy of Down syndrome' was first described relatively recently, in 1984. There remains a paucity of data in the literature about the occurrence of inflammatory arthritis in Down's syndrome; the largest case series for reference is a retrospective chart review of nine children with Trisomy 21 and arthritis, reported in 1990.

Prevalence/Incidence

There are no published population surveys establishing the prevalence and incidence rates of inflammatory arthritis of Down's syndrome. Crude estimates suggest that the incidence of arthritis in children with Down's syndrome is as much as 3 to 6-fold greater than Juvenile Idiopathic Arthritis (JIA) in the general paediatric population. Prevalence has been estimated to be 8.7/1000, compared with the JIA prevalence of 1/1000. Despite these suspected higher incidence and prevalence rates, arthritis is rarely recognised at onset, and is frequently under or misdiagnosed.

Experience of health care professionals in Ireland – setting up a screening programme

The Rheumatology Team at the National Centre for Paediatric Rheumatology (NCPR) in Our Lady's Children's Hospital Crumlin (OLCHC) have experience of caring for many children and young people with inflammatory arthritis of Down's syndrome and have seen first-hand how delayed diagnosis can lead to irreversible joint damage. This joint damage, and consequential functional impairment, is preventable if timely diagnosis and treatment is instigated. We were keen to explore inflammatory arthritis of Down's syndrome in terms of its clinical and radiological features, as well as to investigate the role of established biomarkers in inflammatory arthritis of Down's syndrome.

With the help and support of the members and staff of Down Syndrome Ireland, a national screening programme was set up, offering children with Down's syndrome (aged 0-21 years) the opportunity to attend a local musculoskeletal screening clinic. At this appointment, a medical history was noted, and a full musculoskeletal examination performed. Children with suspected arthritis were invited to attend for Consultant review at the NCPR in OLCHC. At this appointment, diagnosis was confirmed, investigations carried out and management instigated as required.

Findings from the screening programme

To date, just over 550 children and young people with Down syndrome (56% Male) have been screened for inflammatory arthritis of Down's syndrome. A range of musculoskeletal disorders have been detected and documented through the screening

process. After pes planus ("flat feet"), the most common musculoskeletal finding in our cohort of children with Down's syndrome was inflammatory arthritis.

Twenty-two new cases of inflammatory arthritis of Down's syndrome were detected through the screening initiative. Combining those attending the NCPR prior to the start date of the study, and direct referrals throughout the time period of the study, we now have a cohort of 40 cases of inflammatory arthritis of Down's syndrome, the largest reported in the literature to date. Using results from our research, the suspected prevalence of inflammatory arthritis of Down's syndrome is 18-21/1000, greater than previously reported in the literature.

Delay in diagnosis

Comparable to the limited literature available pre-dating our study, we observed a significant delay in diagnosis in our inflammatory arthritis of Down's syndrome cohort (1.7 years, range 0.2-4.9 years) compared with our JIA cohort (0.7 years, range 0.2-2.4 years).

How the condition presents

In terms of disease pattern, children with inflammatory arthritis of Down's syndrome frequently present with a polyarticular arthritis, i.e. five or more joints affected. Small joint involvement of the hands occurs in almost all cases, a feature that appears unique to the arthritis associated with Down's syndrome. Radiological changes were present in 67% of inflammatory arthritis of Down's syndrome cases at diagnosis, 29% had erosions on plain film x-ray, representing irreversible joint damage. These changes were significantly higher than those detected in our JIA comparison group (Radiological changes at diagnosis 24%; Erosions 10%). Again, this is likely due to the significant delay in diagnosis of inflammatory arthritis of Down's syndrome, but could also support the theory that inflammatory arthritis of Down's syndrome is potentially a more aggressive, erosive disease than JIA.

Treatment

Another research finding is that treatment for inflammatory arthritis of Down's syndrome is complicated by drug-associated side effects in a higher proportion of cases when compared to treatment for JIA cases. These findings have allowed us to make evidence-based choices when deciding on appropriate treatment for our patients with inflammatory arthritis of Down's syndrome. This in turn enables improved disease control, minimising irreversible joint damage and leading to better clinical outcomes, whilst ensuring greater patient satisfaction in terms of side effect profile.

An under-recognised condition and the need to raise awareness

Inflammatory arthritis of Down's syndrome is an under-recognised condition that results in chronic disability and functional impairment in a population already at significant risk. Studies of arthritis in Down's syndrome are very limited. Our research, the largest of its kind to date, has shown that there is an increased risk of arthritis in children with T21, with a prevalence double that previously reported. We have found there to be a significant delay in diagnosis, the reasons for this being multifactorial. The disease

pattern appears to be unique to inflammatory arthritis of Down's syndrome, with predominance in the small joints of the hands. Treatment is complicated by a high percentage of drug-associated complications.

To date, our research has increased both knowledge and awareness in the public and professional fields about inflammatory arthritis of Down's syndrome. Children with Down's syndrome and arthritis are now presenting earlier due to the increased awareness about the condition. Similarly, to JIA, timely diagnosis and treatment leads to improved clinical outcomes, which translates to a better quality of life for children with inflammatory arthritis of Down's syndrome.

Current and future research

The investigators are now focusing their research on what might be driving the disease process in inflammatory arthritis of Down's syndrome, by exploring three key areas; immunology, histology and genetics.

1. Immunology – Exploring and measuring the type and number of immune cells in the blood from children with inflammatory arthritis of Down's syndrome and comparing these results to three separate groups of children (children with arthritis without Down's syndrome; children with Down's syndrome and no evidence of arthritis, healthy children without Down's syndrome)
2. Histology – Examining tissue taken from the lining of joints in children with inflammatory arthritis of Down's syndrome and comparing its appearance under a microscope with tissue taken from a child with arthritis without Down's syndrome
3. Genetics – Exploring known arthritis genetic susceptibility markers in children with inflammatory arthritis of Down's syndrome to identify whether they are present and if, like in the arthritis that affects children without Down's syndrome, they contribute to the risk of developing arthritis in children with Down's syndrome.

Identification of differences in immune cells or genetic variants will help improve our understanding of the cause of inflammatory arthritis of Down's syndrome, as well as allow us to develop better diagnostic and therapeutic options. This in turn should lead to marked improvements in clinical care and management of children with inflammatory arthritis of Down's syndrome.

Notes for parents

Consider a diagnosis of arthritis if you notice:

- **A change in your child's behaviour e.g. seeking comfort, irritability, dislikes holding your hand (may suggest arthritis of the fingers or wrist).**
- **Subtle adaptations to overcome difficulty e.g. bum shuffles downstairs.**
- **A change or dis-improvement in handwriting.**
- **Regression in motor milestones.**
- **Your child becomes less active.**
- **Your child walks with a limp.**
- **Your child is slow to get going in the mornings, may suggest early morning stiffness, a sign of arthritis.**
- **Joint swelling, maybe a sign of inflammatory arthritis.**
- **Your child bites their fingers or rubs a particular joint, may suggest arthritis here.**

Notes for Healthcare Professionals

- **Inflammatory arthritis of Down's syndrome is 18-21 times more common than JIA in the general Paediatric population.**
- **Have a high index of suspicion of arthritis when assessing a child with Down's syndrome presenting with change and/or deterioration in function and mobility.**
- **Small joints, wrists and knees are the most commonly affected sites.**
- **Inflammatory arthritis of Down's syndrome may often be insidious and asymptomatic.**
- **A child with inflammatory arthritis of Down's syndrome may present with minimal clinical signs, i.e. joint pain, joint swelling or early morning stiffness.**
- **Look for subtle signs from clinical examination that may suggest a possible diagnosis of inflammatory arthritis of Down's syndrome, e.g. loss of range or loss of hyperextension, especially if there is asymmetry between both sides. This may suggest restrictions from undiagnosed/ untreated inflammatory arthritis of Down's syndrome.**
- **MRI with gadolinium contrast should be the gold standard for definitive diagnosis of inflammatory arthritis of Down's syndrome. Consider if any concerns, as clinically there can be little to aid with diagnosis.**
- **Include a Musculoskeletal Assessment as part of the Annual Surveillance Programme.**

Annual health checks for people with Down's syndrome (aged 14 years plus)

In the past people with learning disabilities have not had equal access to healthcare compared to the general population. This, amongst other reasons, has given rise to poorer mental and physical health and a lower life expectancy for people with learning disabilities. Free annual health checks for adults with learning disabilities, with their GP, were introduced in 2008 as a way to improve people's quality of life.

The annual health check for people with learning disabilities is a Directed Enhanced Service (DES). This is a special service or activity provided by GP practices that has been negotiated nationally. Practices can choose whether or not to provide this service. The Learning Disability DES was introduced to improve healthcare and provide annual health checks for adults on the local authority learning disability register. To participate in this DES, staff from the GP practice need to attend a multi-professional education session run by their local Trust. The GP practice is then paid a sum of money for every annual health check undertaken.

Who can have one?

Annual health checks have been extended to include anyone with learning disabilities aged 14 years or above. So anyone with Down's syndrome aged 14 years or over can have an annual health check.

The benefits of annual health checks

- additional support to get the right healthcare
- increased chance of detecting unmet, unrecognised and potentially treatable health conditions
- action can be taken to address these health needs.

How to get an annual health check

- The GP may get in touch with the person with Down's syndrome to offer an annual health check but this doesn't always happen.
- A person with Down's syndrome and/or a supporter can ask their GP for an annual health check. You do not need to be known to social services to ask for an annual health check.

Not all GPs do annual health checks for people with learning disabilities but they should be able to provide details of other GPs in your area who offer this service.

What happens next?

- The GP practice may send out a pre-check questionnaire to be filled out before the annual health check takes place.
- The GP may arrange for the person with Down's syndrome to have a routine blood test a week or so before the annual health check.

Who attends the annual health check?

If the person with Down's syndrome (age 16 years or over) has capacity and gives their consent, a parent or supporter can attend the health check as well.

How long should an annual health check be?

Guidance from the Royal College of GPs suggests half an hour with your GP and half an hour with the Practice nurse.

What areas of health should be looked at as part of the annual health check?

We have produced a check list for GPs which contains information about what should be included as part of a comprehensive and thorough annual health check. This includes a list of checks that everyone with a learning disability should undergo as part of an annual health check and a list of checks specific to people with Down's syndrome. You can find the health check list at the 'annual health checks' section of our website under 'families and carers and 'health and wellbeing'.

What happens after the annual health check?

Your GP should tell you what they and the nurse have found during the annual health check. You should have a chance to ask any questions you have. Your GP may refer you to specialist services for further tests as appropriate. Your GP should use what they have found during your annual health check to produce a health action plan. This should set out the key actions agreed with you and (where applicable) your parent or carer during the annual health check. Your GP has to do this as part of the annual health check service.

Information about health issues for GPs

There is information at our website for GPs about some of the more common health conditions seen in people with Down's syndrome. You will find this information at the 'annual health checks' section of our website under 'families and carers and 'health and wellbeing'.

GPs learning disability register

People with learning disabilities experience poorer health compared to the rest of the population, but some ill health is preventable. Over one million people in the UK have a learning disability but only 200,000 are on their GPs learning disability register.

We know that people with a learning disability often have difficulties accessing health services and face inequalities in the service they receive. The Government is asking parents and supporters to speak to their GP and ensure their sons/daughters or the people whom they support are registered. It is hoped that this drive will ensure better and more person centered health care for people with learning disabilities.

The Learning Disability Register is a record of people with a learning disability who are registered with each GP practice. The Register is sometimes referred to as the Quality Outcomes Framework (QOF) Register.

If you are not sure you are on the Register, you can ask the receptionist at your GP Practice to check for you.

The doctor may have made a note on the record that a person has Down's syndrome but this does not automatically mean they have been put on the Register. When you speak to the GP about being registered, the needs and support of the person in health settings can be discussed. This information can be entered on the person's Summary Care Record (SCR) so that all health professionals at the practice know about their needs and how best to support them.

If the person is over 16 years of age or older, they must give their consent (see section in this resource about the Mental Capacity Act 2005):

- for information about their support needs to be added to their SCR
- to which information can be shared and with whom

It's never too early (or late) to join your GP's Learning Disability Register; you can join at any age. It's a good idea for children with a learning disability to join the learning disability register at an early age. This means adjustments and support can be put in place before they reach adult services.

Reasonable adjustments in health care

You may have heard of the term 'reasonable adjustments' and wondered what it meant. Since the Disability Discrimination Act (1995) and the Equality Act (2010) (this does not apply to Northern Ireland) public services are required by law to make reasonable adjustments to help remove barriers faced by people with disabilities when trying to use a service. The duty under the Equality Act to make reasonable adjustments applies if you are placed at a substantial disadvantage because of your disability compared to people without a disability or who don't have the same disability as you.

So for people with physical disabilities reasonable adjustments may include changes to the environment like ramps for the ease of wheelchair users. For people with learning disabilities reasonable adjustments may include easy read information, longer

appointments, clearer signs at the practice, help to make decisions, changes to policies, procedures and staff training.

If a patient with Down's syndrome is NOT on their GP's Learning Disability Register then reasonable adjustments to care for that person cannot be anticipated and made.

Authors:

Dr. Orla Killeen, Consultant Paediatric Rheumatologist (Principal Investigator)

Dr. Charlene Foley, Clinical Research Fellow

Dr. Emma Jane Mac Dermott, Consultant Paediatric Rheumatologist

Professor Gerry Wilson, Professor of Rheumatology (Supervisor)

Professor Ursula Fearon, Professor of Rheumatology (Supervisor)

Professor Douglas Veale, Consultant Rheumatologist (Supervisor)

Derek Deely, Clinical Nurse Specialist

Joanna Connolly, Clinical Nurse Specialist

Deirdre Carroll, Clinical Nurse Specialist

Affiliations:

National Centre for Paediatric Rheumatology (NCPR)

Our Lady's Children's Hospital, Crumlin

University College Dublin

Trinity College Dublin

St Vincent's University Hospital, Dublin

The information in this resource is taken from an article published in DSA Journal 138 Autumn/Winter 2018

Publication date 2020

The Down's Syndrome Association provides information and support on all aspects of living with Down's syndrome.

We also work to champion the rights of people with Down's syndrome, by campaigning for change and challenging discrimination.

A wide range of Down's Syndrome Association publications can be downloaded free of charge from our website.

Contact us

Down's Syndrome Association

Langdon Down Centre,
2a Langdon Park, Teddington,
Middlesex, TW11 9PS

t. 0333 1212 300

f. 020 8614 5127

e. info@downs-syndrome.org.uk

e. training@downs-syndrome.org.uk

w. www.downs-syndrome.org.uk

Wales

e. wales@downs-syndrome.org.uk

Northern Ireland

e. enquiriesni@downs-syndrome.org.uk



WorkFit® is a registered trade mark of the Down's Syndrome Association. Photographs courtesy of Monkey Business/Gino Santa Maria/Denis Kuvaev www.shutterstock.com



www.dsactive.org



www.langdondownmuseum.org.uk



www.langdondowncentre.org.uk

© Down's Syndrome Association 2020