

**STICKLER SYNDROME
SUPPORT GROUP
(SSSG)
Registered Charity: 1060421**

**JOINT INVOLVEMENT WITHIN
STICKLER SYNDROME**

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1. AN OUTLINE OF THE PROBLEMS

- The degree of joint involvement within Stickler Syndrome is extremely variable. Some people may experience few joint problems, whilst in others the problems are far more significant. The features commonly seen in children with Stickler Syndrome are loose jointedness (double-jointedness or hypermobility) and prominent joints (e.g. knees, finger joints).
- If bone problems are present at birth, then talipes equinovarus or club foot can occur. This is a condition where the foot is twisted downwards and inwards so that the patient walks on the outer edge of the front of the foot. It is easy to correct by orthopaedic splinting in the early months of infancy.
- Later in life the joints may become stiff, and early onset of osteoarthritis can occur. Osteoarthritis, which is common in an ageing population, can occur at a much earlier age in people with Stickler Syndrome (e.g. early 30s and 40s). Changes similar to osteoarthritis can affect the neck and back and are referred to as cervical and lumbar spondylosis respectively. However, not all people with Stickler Syndrome will develop these changes.
- The symptoms affecting the joints (e.g. pain, stiffness) can be aggravated by excessive overuse. In childhood the features may mimic (and sometimes be confused with) other forms of juvenile arthritis, or other conditions associated with hypermobility, such as Osteogenesis Imperfecta (OI), Marfan Syndrome and Ehler-Danlos Syndrome (EDS).
- X-ray changes affecting the spine include changes that resemble 'spondylo-epiphyseal dysplasia' which refers to irregularity of the bones of the back (vertebral bodies). These changes can be incidental and cause no symptoms but are helpful in making a diagnosis. Other x-ray changes of dysplasia (irregularity of the shape of

the bone) can be seen to affect the ends of the femur (thigh bone) or other long bones.

- Hypermobility, or increased mobility of certain joints, can cause the joints to become painful and prone to dislocation. The kneecap, shoulder or even the hip may dislocate, causing pain, although they can usually be returned to the correct position. Children are also prone to dislocated joints, and parents, school teachers, youth leaders, etc should be aware of this potential problem when a child is playing and exercising.
- Hypermobile joints caused by an altered collagen protein in Stickler Syndrome can also lead to hernias or varicose veins. A flat arch to the foot may be present and this leads to foot ache, particularly when standing for a long period.

2. FOUR MAIN CAUSES OF HYPERMOBILITY

- The shape of the end of the bones, like the shoulder and the hip (where they move at the joints) is important as joints normally have a large range of movement. If a shallow rather than a deep socket is inherited, there will be a large range of movement in these particular joints, which means they can dislocate more easily.
- Problems with collagen or other proteins will cause weak or stretched ligaments (the bands that hold the joints in place). These proteins include elastin, which gives elasticity, and collagen, which gives strength. The collagen fibres in the ligaments tend to bind together more as we get older, which is one reason why we become stiff with age.
- The tone of the muscles will affect how the joints are held – loosely or rigidly. The tone, or stiffness, of muscles is regulated by the nervous system, which in turn controls the range of movement.
- The sense of joint position and movement is difficult to describe, as it is the inner sense that tells you exactly

where the joint is positioned and whether it is overstretched. Close your eyes and try to describe the exact position of your joints. If you can't, you may be more likely to overstretch a joint before you notice you have done so.

Some people may need to modify their way of life in order to protect the joints and prevent complications, such as the onset of early osteoarthritis, which can be the result of overuse of loose joints. Like all degenerative conditions, as the cartilage covering the end of the bone wears away, leaving bone rubbing on bone, the joints become stiff, painful and difficult to move. The most common joints affected are the hips, knees, ankles, neck, back, hands and feet. Walking with an affected hip joint can be difficult and the hip will gradually alter in shape. In severe cases, this can result in the leg on that side becoming noticeably shorter than the other. Osteoarthritis of the knees can also cause a wide range of deformities. It can make the knee look knobby and the affected person may appear to be bow-legged or knocked-kneed. Some people will have difficulty walking up and down stairs, and it is possible to hear creaking and grating noises whenever the arthritic joint is moved. The most common joint in the foot to be affected by osteoarthritis is the joint at the base of the big toe. Wearing comfortable shoes can help to relieve the pressure on the joint.

3. EXAMINATION AND TESTS

Tests or investigations are often used for one of the following reasons;

- to make a diagnosis
- to rule out other explanations for your symptoms
- to assess how active your arthritis is.

- to predict how your arthritis will develop.
- to work out the best form of treatment.
- to monitor drug therapy.

The diagnosis is based on your clinical history – what you have told the doctor and what the doctor has found by examining you.

3.1. BLOOD TESTS

Full Blood Count (FBC) - this will help the doctor to decide whether you are anaemic (lacking haemoglobin), and whether you have normal white cells (that fight infections and normal platelets (which clot the blood). People who have arthritis often have minor abnormalities of the full blood count. It can also be affected by some drug therapies.

3.2. ERYTHROCYTE SEDIMENTATION RATE (ESR)

This simple test measures how quickly the red blood cells settle when left to stand in a tube. This will reflect how much inflammation your arthritis has caused, and is a good measure of whether your arthritis has improved or worsened.

3.3. URATE

We all have urate in our blood. It is a waste product from the normal breakdown of protein and comes out in the urine. If the level builds up then some of it solidifies as crystals, particularly in the joints of the hands and feet. These crystals can cause severe inflammation, which we call gout. The level in the blood can help to make the diagnosis of gout, and can also be used to monitor the response to treatment.

4. XRAY

This is the most tried and tested way to get a picture of your bones and joints. These are done in an x-ray

department by a radiographer, who will explain to you which part of your body is being x-rayed. Patients who attend a rheumatologist for the first time may have a routine chest x-ray, because chest diseases are sometimes associated with arthritis. To make the image, x-rays are passed through your body and captured on an x-ray film which is then developed and examined. They are particularly good at showing abnormalities of the bone, but rarely show problems with other tissues nor show early arthritis very well. However they can help a doctor to diagnose arthritis through damaged areas of bone. Abnormalities caused by aging are frequently seen on x-rays of the spine, even in patients without pain. Conversely normal looking x-rays may be developed despite severe pain from inflammation in or around the joints. In addition x-rays can show apparent damage or abnormalities which may not be the cause of the pain, swelling or stiffness. Therefore your doctor will not rely on x-rays alone, but will interpret the images alongside findings from the physical examination and from other tests.

5. OTHER METHODS OF IMAGING

There are other ways of scanning the body to produce images and other ways of using x-rays, as well as the standard form of x-ray.

5.1. ARTHROGRAPHY

This uses a dye which is injected into the joint to image the joint so that it will show up in more detail on the x-ray. Occasionally dye can be injected into a blood vessels to access the circulation under X-ray, and this is called an arteriogram or a venogram.

5.2. COMPUTERISED TOMOGRAPHY (CT)

This uses x-rays which record images of sections or 'slices' of the body. These multiple images are then

transformed by a computer into cross-sectioned pictures. CT scans can give a detailed picture of the skeleton, but can also show other types of tissue, such as the muscles, which cannot be seen on an ordinary x-ray.

5.3. ISOTOPE BONE SCAN

An isotope is a chemical which gives off a type of radioactivity called gamma rays. A small amount of which can build up a picture showing which areas of the bone are inflamed. There is no need to worry, as the dose of radiation is very small and will quickly be removed from the body in the urine.

5.4. MAGNETIC RESONANCE IMAGING (MRI)

This uses high frequency radio waves in a strong magnetic field. Radio-waves interact harmlessly with the water molecules in the body's tissues, and the signals which come back are processed by a computer to build up pictures of the inside of the body. An MRI scan produces pictures of the soft tissues – particularly cartilage, tendons and nerves – that cannot be detected by x-rays. MRI scans are often used to detect early or minor abnormalities in these soft tissues. They can also show up signs of inflammation and are particularly useful for the spine and knee. MRI scans can show numerous differences between individuals, many of which are completely normal, therefore it is important that the results are interpreted by the doctor who has ordered the scan.

6. OTHER EXAMINING TECHNIQUES

6.1. SYNOVIAL FLUID ANALYSIS

All normal joints have a small amount of fluid in them to act as lubrication. If this fluid builds up it can make the joint swell. Examining this fluid can be very important in diagnosing infections or gout. For examination, the fluid is taken from the joint using a needle and syringe. The

needle does not go into the bone, but into the space or cavity between the bones. This is often no more uncomfortable than having a blood test. Removing the fluid can also help to reduce discomfort and swelling. The fluid can then be examined in the laboratory. It will usually be analysed for colour and clarity, and may be examined under a microscope to check for blood cells, crystals and bacteria.

6.2. BIOPSY

A biopsy is when a small piece of tissue is removed from the body so it can be examined under the microscope. Virtually any body tissue can be biopsied and can be done under a general or local anaesthetic. It will be done when it is absolutely necessary to make a diagnosis. Some forms of arthritis can affect many parts of the body, and it is essential to examine the tissue under the microscope to make a proper diagnosis and work out the appropriate treatment.

7. TREATMENTS

7.1. PAIN KILLERS

Such as paracetamol are prescribed to help a patient to manage his or her life, but should generally not exceed the recommended dosage.

7.2. NSAIDS

Non steroidal anti-inflammatory drugs - can relieve mild inflammation and stiffness, but they have no long-term effect on the disease. They are often taken in addition to simple painkillers but you should discuss their use with your GP or rheumatologist before you start to take them routinely as they have certain side effects if taken for prolonged periods. As with all pharmaceutical preparations a patient may need to try several before finding one that is suitable for his or her needs. There are many different

drugs on the markets, and those most commonly used are Ibuprofen, diclofenac, naproxen, indometacin and ketoprofen. They should be taken with or shortly after food and as directed by your doctor. You should also take a full glass of fluid with NSAID tablets. Some are taken once a day, and are known as slow release, sustained release or retard. Others are taken 2-3 times a day. However these drugs are not suitable for all patients, particularly those with other medical problems, and may not be prescribed.

7.3. CORTISONE INJECTIONS

Short term relief from symptoms of moderate osteoarthritis of the knee may be obtained from a cortisone injection, particularly if there is some inflammation or fluid in the joint. This is usually recommended under specialist care in conjunction with other treatments and may not be suitable in all cases.

7.4. CONNECTIVE TISSUE MANIPULATION

Some Stickler Syndrome patients have found connective tissue manipulation beneficial, although this technique is not readily available at all physiotherapy centres. Connective Tissue Manipulation is performed by a physiotherapist using the soft pads of the fingers to move one layer of skin on the layer below. The movement creates a short stretch, reflex, thus creating an impulse, which spreads out through the connective tissue. Benefits are not immediate and it will not work for everyone. The treatment is intense and tiring for both the patient and the physiotherapist, but it is not painful, just uncomfortable. Your GP or rheumatologist can advise.

7.5. ARTHROSCOPY

Some individuals (e.g. those with intermittent pain and locking of the knee joint due to 'loose bodies' in the joint) may undergo a 'keyhole' procedure to wash out the knee or to remove any loose bodies. A small telescope is used

to look inside the joint, and also can be used to directly access the joint to perform a biopsy, or to perform surgery such as the removal of a torn meniscus (cartilage). Depending on what is required this will be performed either under a local or general anaesthetic.

7.6. JOINT REPLACEMENT SURGERY

Those with painful, significant osteoarthritis may ultimately be considered for joint replacement surgery. The most common operations in Stickler Syndrome patients involve the hip or knee. Specialist orthopaedic centres will usually be involved for younger patients, people with other joint involvement problems or those needing surgery to correct serious joint deformity.

8. SELF HELP EXERCISES

Painful joints may be helped by some or all of the following:

- Weight loss in obese individuals can significantly help relieve knee joint symptoms.
- At home a warm bath or a heated pillow can relieve pain.
- Active treatments including gentle exercises may help to strengthen the muscles surrounding the affected joints, or improve posture. However no-one should embark on a course of exercises without discussing their plan with a GP or a rheumatologist who knows about Stickler Syndrome.
- In some cases hydrotherapy in a pool may be beneficial, but is not always available locally within the NHS.
- A technique called *Transcutaneous Electrical Nerve Stimulation* (TENS) may help. This is a method of electrical stimulation which primarily aims to provide a degree of pain relief and can be particularly helpful for

back pain. This is non invasive and has few side effects when compared with drug therapy, although about 2-3% of patients complain of an allergic type skin reaction. This is almost always due to the material of the electrodes (the conductive gel or the tape used to hold the electrodes in place). These pocket machines can be purchased at any good chemists, but patients should discuss beforehand whether they should use one of these machines with their GP, rheumatologist or physiotherapist.

- Acupuncture has proved affective for relieving musculoskeletal pain in some Stickler Syndrome patients. Again the GP, rheumatologist or physiotherapist will be able to advise whether this is a suitable course of action.
- A course of physiotherapy may help. A collar may help a painful neck. Ultrasound or other treatments using sound waves, laser and magnetic wave therapy may also reduce pain and swelling of joints. A GP, rheumatologist or physiotherapist can advise depending on the patient's individual needs. Some patients find that hot paraffin wax treatment helps finger stiffness.
- Gentle regular exercise can help joints and will allow the joints natural range of movement, help them to remain supple and mobile without abnormal strain. The simplest exercise of course is walking, but if the patient also had a visual impairment it isn't a simple matter of taking oneself off for a brisk walk.
- Most physiotherapists can offer printed instructions on gentle exercises, but if there is also a retinal problem, do check with your physiotherapist and ophthalmic surgeon that the exercises are appropriate for you.

9. GENERAL EXERCISES

Before embarking on these exercises you will need an upright chair, but always remember that if you cannot

make a movement, **DON'T FORCE YOURSELF**. Exercises are designed to help, not to punish, and never strain yourself.

- Sitting on the chair and keeping feet on the ground, lift up your heels and toes. Do this with each foot, then with both feet at the same time.
- For neck flexibility. Sit comfortably in the chair and let the head sag forward, but do not push, then lift it up slowly. Let your head fall gently to the side, then bring it up again. Do this for both sides.
- Place your bottom against the back of the chair. With your spine touching the chair back all the way up; draw yourself up – chest out, head up. Relax and repeat.
- Sit comfortably in the chair. Drop your head forward until it touches the chest. Straighten up again.

10. SPINE AND BACK EXERCISES

- Stand up straight. Bend forwards towards your toes as far as you can. Then straighten up again, folding your arms across your chest, lean backwards so that you are looking towards the ceiling. Then bend forwards again. This exercise should be done as a gentle, continuous curling, starting at the base of the spine and working upward.
- Stand up straight. Bend slowly, sideways, running your left hand down your left thigh. Then straighten up again and do the same with the right.
- Stand up straight. Put your hands on your hips and, without moving your feet, rotate the spine to turn first to the right and then to the left.
- Lie as flat as possible on a bed, and press the head and shoulders hard back into the bed. Relax and repeat.
- Lie as flat as possible. Using your tummy muscles, lift your head up to look at the feet.

- Sitting in an upright chair, lean backwards arching your back whilst taking a deep breath in. Relax and breathe out. Repeat.
- Sit up straight in a chair and twist the body round to look behind, keeping your bottom squarely on the chair. Repeat the exercise turning the other way.
- Sit upright in the chair. Lean forward to put the head on the knees, straighten to the upright position slowly.

11. FEET AND ANKLE EXERCISES

- Sit upright with legs crossed and pull feet up and down from the ankle. When pushing down point the toes hard towards the floor. Repeat with the other foot.
- Sit upright and circle the foot round from the ankle drawing a large circle in the air with the big toe. Repeat with the other foot.
- Sit upright and press the toes flat against the floor and, keeping them straight raise the ball of the foot off the ground to make a bridge. This is a very important exercise to re-train the small 'sling' muscles supporting the arches of the foot. Repeat with the other foot.

12. EXERCISES FOR THE KNEES

- Lie on your back with your knees outstretched. Slowly bend up one knee as far as it will go, then stretch the leg out straight again. Then do the same with the other leg.
- Lie straight with your feet pointing upwards. Slowly lift one leg – not very high – but just enough so that it is not touching the bed. Keeping your knee straight and your thigh muscles braced, lower it again slowly until the calf touches the bed and then relax. Repeat the exercise with each leg in turn.

13. SHOULDER EXERCISES

- Sit on an upright dining chair. Let one arm hang down by the side and swing it forwards and backwards in a pendulum movement. Repeat with the other arm.
- With the arm hanging as before, swing it away from the side.
- Support the hand and wrist with your other hand; raise the elbow away from the side.
- Lift and circle the shoulders, first one way and then the other trying each time for the maximum mobility.

All these exercises should be done for short periods, with a rest between, and preferably when you are warm from a bath or shower.

14. HOW YOU CAN REACH US

Write to:

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