

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

UK ADULT DATA

STICKLER SYNDROME QUESTIONNAIRE SURVEY

November 1999

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UK ADULT DATA

- ◆ Data was gathered from 79 adults (aged 17 and over) living in the United Kingdom.
- ◆ 31 (39%) respondents were male; 48 (61%) were female.

GENERAL

Diagnosed as having Stickler syndrome by a medical professional

Yes	95%
No	5%

Data from the four respondents without a clinical diagnosis of Stickler syndrome has been included on the basis that all four showed symptoms of Stickler syndrome and had one or more close relative who had been clinically diagnosed with the condition.

Age at diagnosis

Aged 0-10	3%
Aged 11-20	19%
Aged 21-30	19%
Aged 31-40	25%
Aged 41-50	12%
Aged 51-60	8%
Aged 61+	4%

Average age of respondents	40.6 years
Average age at diagnosis	32.5 years

By whom first diagnosed

Ophthalmologist	73%
Geneticist	24%
Other:	Optician (1) Neurologist (1)

Diagnosis by ophthalmologist at first retinal detachment

77% of UK adults who had experienced one or more retinal detachment had been diagnosed by an ophthalmologist. 37.5% of these adults reported the same age at diagnosis and age of first retinal detachment, leading us to conclude that the initial detachment had provided the basis for a diagnosis.

However, 26% of adults who had experienced one or more retinal detachment and been diagnosed by an ophthalmologist were not diagnosed at age of first retinal detachment. The majority of these had had their first detachment during the 1980s or earlier when awareness of Stickler syndrome was low. 7% were not diagnosed by an ophthalmologist until between 30 and 44 years after their first detachment but 4% reported diagnosis by an ophthalmologist *before* their first retinal detachment.

Height

Males (n=31)

5' (1.52m) - 5'5" (1.65m)	10%
5'6" (1.68m) - 5'11" (1.80m)	32%
6' (1.83m) or above	58%

Average height 5'9" (1.75m)

Females (n=46)

4'5" (1.35m) or below	4.5%
4'6 (1.37m) - 4'11" (1.5m)	13%
5' (1.52m) - 5'5" (1.65m)	37%
5'6" (1.68m) - 5'11" (1.80m)	41%
6' (1.83m) or above	4.5%

Average height 5'2" (1.58m)

Weight

Males (n=30)

Lowest weight	7st 7lbs (48kg)
Highest weight	18st (114.5kg)

Average weight 12st 6lbs (79kg)

Females (n=46)

Highest weight	14st 6lbs (95kg)
Lowest weight	6st 1lb (39kg)

Average weight 10st 11lbs (69kg)

Genetic counselling

24% answered 'yes' to the question "Have you had genetic counselling for Stickler syndrome?" However, the term 'genetic counselling' was not defined, so we cannot comment on the nature nor duration of the counselling offered to our respondents.

Genetic testing

24% reported that they had had DNA screening or genetic testing for Stickler syndrome (only a third of these had also reported receiving genetic counselling). COL2A1 had been implicated in 26% of those tested, with one further respondent expecting DNA results to confirm COL2A1 based on clinical assessment. COL11A1 was named by one respondent, with a further two respondents commenting that the faulty gene was "the one which is linked to type 2". 42% of those tested did not know which gene was responsible, two adding, "Not informed - blood frozen for testing later". Several appeared unsure about whether they were supposed to receive results or not. When asked which gene was implicated, one respondent wrote, "COL2A1? Forgotten - told verbally in 1991/2."

Relatives with Stickler syndrome

73% reported relatives (living or deceased) with Stickler syndrome.

EYES (n=79)

Percentage of sample first diagnosed by an ophthalmologist 73%

Current vision:

Myopic	76%
Vision in both eyes	62%
Vision in one eye only	32%
Totally blind	6%

Registration:

Registered as partially sighted	15%
Registered as blind	16.5%

Retinal detachment:

Have experienced retinal detachment	61%
Number of detachments per adult:	
1 detachment	23%
2 detachments	23%
3 detachments	23%
4 detachments	10%
5 detachments	8%
6 detachments	4%
7 or more detachments	6%
Average age of first detachment	25.5 years
Vitrectomy	52%
(of whom 20% have had a vitrectomy in both eyes)	

Awareness of risk of retinal detachment and need to seek medical help immediately:

- ◆ Stickler syndrome diagnosed before first retinal detachment; sought medical help immediately 12.5%
[N.B. However, only half of the above knew of the high risk of retinal detachment within Stickler syndrome.]
- ◆ Stickler syndrome undiagnosed at first retinal detachment, but sought medical help immediately 81%
- ◆ Did not recognise symptoms of retinal detachment nor seek medical help immediately 23%
- ◆ Would now recognise the symptoms and seek medical help immediately 94%

Cryotherapy and laser treatment:

Cryotherapy treatment to the retina	12%
(of whom: cryotherapy as part of surgery for retinal detachment	20%
cryotherapy as a preventive measure	40%
cryo. as part of surgery AND as a preventive measure	40%)
Laser treatment to the retina	22%
(of whom: laser as part of surgery for retinal detachment	19%
laser as a preventive measure	44%
laser for glaucoma	6%
laser as part of surgery AND as a preventive measure	25%
laser as part of surgery and for glaucoma	6%)
Cryotherapy AND laser treatment	28%

Glaucoma:

Have glaucoma 20%

(of whom 44% have glaucoma in both eyes)

Glaucoma treated with:

Eye drops 100%

Tablets 31%

Surgery 44%

[N.B. Many respondents reported more than one form of treatment for glaucoma.]

Cataract:

Have (or have had) a cataract 65%

(of whom: bilateral cataracts 69%

both cataracts removed 29%

one cataract removed 41%

no cataract removed 29%)

Lens implant following cataract surgery 69%

Awareness of family implications:

Advised to have children or siblings examined by an ophthalmologist 59.5%

Hospitals currently attended for eye care/treatment:

Moorfields Eye Hospital, London 23%
Addenbrooke's NHS Trust, Cambridge 16.5%
Manchester Royal Eye Hospital, Manchester 8%
Royal Berkshire Hospital, Reading 6%
Aberdeen Royal Infirmary, Aberdeen 5%
Birmingham and Midland Eye Centre, City Hospital, Birmingham 5%
Walsgrave NHS Trust, Coventry 3%
Luton and Dunstable Hospital, Luton 3%
West Norwich Hospital, Norwich 3%
Princess Alexandra Eye Pavilion, Edinburgh 3%

Hospitals cited once only:

England Blackburn Royal Infirmary, Blackburn
Sunderland Eye Infirmary, Sunderland
Leeds General Infirmary, Leeds
Bradford Royal Infirmary, Bradford
West Suffolk Hospital, Bury St. Edmunds
BUPA Dunedin, Reading
St. George's Hospital, London
St. Thomas', London
Frimley Park Hospital, Surrey
Mayday, Thornton Heath, Surrey
Maidstone Ophthalmic, Maidstone
Southampton Eye Hospital, Southampton
Wales Singleton Hospital, Swansea
Scotland St. John's, Livingston, West Lothian
Gartnavel General Hospital, Glasgow
Eire The Eye Clinic, Dublin

Six respondents who named a hospital added that they had been discharged and told to go back if they experienced any problems. Three (one of whom is totally blind) stated that they did not attend any hospital for eye care or treatment. One

gave 'Boots Opticians' as the source of current eye care and treatment. One was seen by an ophthalmologist at a local surgery.

Other comments and eye problems:

Treatment-related comments:

- ◆ 'Other glaucoma treatments include trabeculectomy (c. age 33) which was unsuccessful, and cyclocryotherapy. Allergic to preservative in eye drops.'
- ◆ 'Cataracts developed as a result of surgery for detachments and follow-up surgery. The lens implant slipped in left eye and detached the retina for a second time.'
- ◆ 'Rejection of implants/scleral buckle breaching on several occasions requiring surgery to reinsert and restitch scleral buckle.'
- ◆ 'Coping with double vision following retinal detachments became my biggest problem. The hospital was pleased with the success of the retinal operation but reluctant to deal with diplopia - didn't want to upset the status quo!'
- ◆ 'I had a bad reaction to my vitrectomy in my right eye so the replacement liquid was removed. The supporting strap in my right eye was unstable for a period thereby wearing away the surrounding membranes as it moved around and this has caused the right eye membrane to become very thin.'
- ◆ 'Had a short-term increase in pressure in my left eye following my second unsuccessful retinal detachment operation through incorrectly prescribed eye drops. Pressure decreased almost immediately when different drops prescribed. This was in 1973/4 - cannot remember the name of the eye drops concerned.'
- ◆ 'I had three operations on my eye because one implant is floating at the back of my left eye.'
- ◆ 'Because I was relatively young when my cataracts were removed, the scar tissue formed 'pearls' on both eyes. I therefore had to have a hole cut through these with a laser to enable me to see again. I was registered, but after several operations, my sight improved.'
- ◆ 'Was myopic when young, but not now because of lens implant.'

Enucleation:

- ◆ 'Left eye enucleated following retinal detachment.'
- ◆ 'I have an artificial eye from when I lost my vision after retinal detachment.'
- ◆ 'Did have glaucoma in the right eye, but had the eye removed.'

Other problems identified by respondents:

- ◆ 'Throbbing pain as if swollen - not thought of as a problem or recognised by opticians.'
- ◆ 'Do not have glaucoma but pressure fluctuates. Intermittent/periodic pain, bloodshot, swollen. Retinal cysts, scarring. Numerous small bleeds.'
- ◆ 'Vision hopeless at night.'
- ◆ 'Difficulty seeing in the dark.'
- ◆ 'Pain, redness and soreness in left eye. Vision restricted by strong sunlight.'
- ◆ 'Both eyes very oversensitive to bright light. Has been present in some degree for many years - acute after cryotherapy and has never returned to same level as before the cryo. I need to wear sunglasses a lot of the time now. My consultant explained that this oversensitivity to light is caused by lack of sufficient pigment in the retinas causing light to scatter, be reflected and cause more glare within the eyes. I was very grateful for the explanation.'

- ◆ 'Latterly haemorrhages in left eye leading to total blindness.'
- ◆ 'Have lost my colour vision.'
- ◆ 'Dry eyes for which I have to put in drops for artificial tears.'
- ◆ 'Although my eyesight is now corrected with glasses, I still find my eyes tire easily - also suffer from dryness in my left eye since surgery.'
- ◆ 'I was born with a cast in the left eye which was treated by surgery at the age of 3 years. That eye was always considered to be a 'lazy' eye.'
- ◆ 'I have lazy eyes and both long and short-sighted problems.'
- ◆ 'My pupil in my right eye is not very quick to focus.'

Additional eye disorders diagnosed:

- ◆ 'In my family, we have the additional feature of retinitis pigmentosa and therefore additional eye problems.'
- ◆ 'Cataracts were congenital. Thought to be in addition to Stickler syndrome as non-Stickler parent also had congenital cataracts. Also have nystagmus in both eyes.'

MOUTH AND FACE (n=79)

Percentage of sample first diagnosed by a cleft surgeon 0%

Cleft palate:

Born with a cleft palate 23%

Type of cleft:

Submucous 33%

Other 66%

Respondents ticking 'Other' were then asked to describe their cleft in their own words. Responses can be summarised as follows:

Don't know/not sure 58%

Hard and soft palate 25%

Soft palate only 17%

Other terms used by respondents in connection with their cleft included 'Bilateral' and 'Full'. Those answering 'Don't know/not sure' might have been better able to indicate the nature of their cleft if they had been given a list of standard descriptors.

Age of diagnosis of cleft:

At birth 67%

Neonatally 11%

Age 3 years and above 11%

[N.B. 'Neonatal' indicates that diagnosis occurred between 1 and 14 days after the birth. The two respondents whose clefts were diagnosed after the age of 3 - one at age 14, and one at age 27 - both had submucous cleft palates.]

Age at which cleft repaired:

6 months and under 17%

7-12 months 28%

13-18 months 22%

19 months and over 11%

[N.B. Two respondents gave two ages for cleft repair, but only the age of first repair has been included in the above figures.]

Of the three early cleft repairs, one gave age of repair as 1-2 days, one gave 5 months (and again at 7 months) and one gave 6 months. Of the cleft repairs on older children, one respondent indicated that repair was carried out at age 3 (and again at age 12), and one reported repair at age 7.

Cleft not repaired 22%
(of whom 75% had a submucous cleft palate).

Pierre Robin sequence:

Diagnosed as having Pierre Robin sequence	10%
Age at which diagnosis changed to Stickler syndrome:	
Aged 10 and under	12%
Aged 11-20	37%
Aged 21-30	50%
Average age of change of diagnosis from Pierre Robin sequence to Stickler syndrome	19 years

Feeding, breathing and speech difficulties in childhood:

Had problems feeding as a baby	24%
(of whom 74% had been born with a cleft palate)	
Had problems breathing as a baby	11%
(of whom 78% had been born with a cleft palate)	
Had problems with speech as a child	29%
(of whom 56% had been born with a cleft palate)	
Had speech/language therapy as a child	21%
(of whom 59% had been born with a cleft palate)	
Had problems with speech as an adult	14%
(of whom 64% had been born with a cleft palate, and 91% had reported speech problems as a child).	

Oral/facial features:

High-arched palate	33%
Bifid uvula	8%
Flat midface with a broad and flat nasal bridge (<i>midfacial hypoplasia</i>)	40%
Prominent eyes	19%
Small vertical fold of skin from each eyelid down across the inner corner of the eye (<i>epicanthic folds</i>)	15%
Short bottom jaw (<i>micrognathia</i>)	33%
Teeth which do not bite together well (<i>malocclusion</i>)	38%

Surgery and/or treatment to the mouth or face:

Pharyngoplasty	4%
Nasal/midface bone or cartilage grafts	5%
Orthodontic treatment	30%
Surgery on the upper or lower jaw	4%

Other comments and problems with the mouth or face:

Other surgery/treatment:

- ◆ 'Had surgery because tongue-tied. Still very short under the tongue where they clipped it. Also had tonsils out because I couldn't talk very well (they later found out I was deaf).'
- ◆ 'Nasal bone punched and scraped on numerous occasions between ages of 10-14 years. Tonsils and adenoids removed at 18 months to try and help breathing and speech.'
- ◆ 'Constant infections; removal of tonsils etc. at age 4/5.'
- ◆ 'Had a lot of dental problems due to a small jaw. Top four teeth at front removed at 16. Impacted wisdom - went into hospital for removal.'
- ◆ 'As a child I had to have five teeth removed and a brace fitted as I had too many teeth for my mouth and the adult teeth weren't growing through straight.'

- ◆ 'I had four or more teeth removed to improve my teeth.'
- ◆ 'I had orthodontic surgery at the age of 18 to push top teeth back and had to wear a brace to widen upper palate for about a year before the operation.'
- ◆ 'Micrognathia and malocclusion were not diagnosed in me until I was an adult. Orthodontic treatment followed on after the initial diagnosis.'
- ◆ 'Malocclusion corrected by dental treatment in my early teens (brace).'
- ◆ 'Orthodontic treatment improved malocclusion.'
- ◆ 'Didn't have orthodontic treatment but it was recommended as a child.'

Speech problems:

- ◆ 'I had and continue to have a lisp.'
- ◆ 'Very conscious of speech - find certain words difficult to pronounce. Words tend to tumble out when I am tired, sometimes in the wrong order!'
- ◆ 'I have to be careful when speaking as my mouth fills with saliva and nearly chokes me. My words get mixed up.'
- ◆ 'Speech therapy assessment as a child but no treatment.'

Other oral/facial problems:

- ◆ 'In later life I have experienced pain in the jaw and facial bones/joints. This varies and has progressed in phases over the years. Originally I thought it was a chain reaction from the pain and mechanical problems with my spine and especially my neck. It has progressed through to my jaw, cranial and facial bones. At times they grate on each other and cause acute pain. I experience pain in many places around my head including the bones around my ears, eyes, nose and mouth.'
- ◆ 'Roof of mouth often painful, tender. Sinus problems - rhinitis /sinusitis.'
- ◆ 'I have had problems with too many teeth in both jaws.'
- ◆ 'Maleruption of teeth.'
- ◆ 'Lost all teeth early twenties. Wear denture, for years now only at top as not enough for denture to sit on in bottom half of mouth.'
- ◆ 'Don't know if relevant, but I have fairly soft teeth - they break easily.'
- ◆ 'Gums bleeding (pyorrhoea).'
- ◆ 'Constant mouth ulcers.'

Improvement of appearance with age:

- ◆ 'Had prominent eyes as a baby.'
- ◆ 'Had flat midface as a child but no problem now.'
- ◆ 'Had these problems as a baby and child but they have ended with age.'

HEARING (n=79)

Percentage of sample first diagnosed by an audiologist 0%

Ear infections:

As a child:

Frequent ear infections 25%

Diagnosed with 'glue ear' (*otitis media*) 13%

(of whom 30% were treated by antibiotics only, 30% by the insertion of grommets only, and 30% by both antibiotics and grommets. 10% recorded no treatment.)

As an adult:

Frequent ear infections 9%

(of whom 86% also reported frequent ear infections in childhood, and 57% reported being hard of hearing as an adult)

'Glue ear' 3%

Hearing loss:

Hard of hearing 38%

Hearing loss in one ear only 13%

(of whom: mild 75%
severe 25%)

Bilateral hearing loss 87%

(of whom: bilateral mild 19%
bilateral moderate 34%
bilateral severe 19%
bilateral profound 4%
mild/moderate 8%
mild/severe 8%
moderate/severe 4%
severe/profound 4%)

Nature of hearing loss:

Conductive only 0%

Sensorineural only 27%

Conductive *and* sensorineural 27%

Don't know 37%

Age when hearing loss first diagnosed:

Aged 0-10 40%

Aged 11-20 10%

Aged 21-30 7%

Aged 31-40 3%

Aged 41-50 7%

Aged 51-60 3%

Aged 61+ 3%

Hearing loss worse now than when first diagnosed 60%

[*N.B.* It is not clear from our survey whether this deterioration was felt subjectively, or whether an actual decrease in hearing levels had been recorded since diagnosis.]

Tinnitus:

Suffer from tinnitus		23%
In one ear only		33%
(of whom: mild		33%
moderate		66%)
In both ears		61%
(of whom: mild		91%
moderate		9%)
In head		50%

When asked to describe the noises heard:

- ◆ 'A high-pitched whistle.'
- ◆ 'Whistling.'
- ◆ 'Smooth rushing sound.'
- ◆ 'Low buzz and crackles.'
- ◆ 'Like listening to a conch shell. Musical - like pipes.'
- ◆ 'High-pitched ringing noise when everything else is quiet.'
- ◆ 'Like a high-pitched radio signal.'
- ◆ 'Humming like a tuning fork, buzzing.'
- ◆ 'Humming noises.'
- ◆ 'Buzzing or frying sound.'
- ◆ 'Buzzing noises.'
- ◆ 'Like a pressure cooker letting off steam and hissing.'
- ◆ 'Airplane and motorbike.'
- ◆ 'Ringing and airplane noises.'
- ◆ 'Ringing or buzzing noise.'
- ◆ 'Ringing/buzzing.'
- ◆ 'Generator.'

Hearing aids:

Wear a hearing aid for diagnosed hearing loss	47%
(of whom: In one ear only	43%
Bilateral	57%)

Value of hearing aid(s) as perceived by the wearer:

Help(s) a lot	21%
Help(s), but only in certain situations	71%
Do(es) not help at all	7%

(N.B. The latter reported profound loss: 'Born deaf'.)

Other comments and problems with the ears and hearing:

Comments relating to surgery/treatment:

- ◆ 'In November '94 I had a radical mastoidectomy on my left ear, since which my hearing loss has deteriorated but I have less frequent ear infections. I have to have regular 'vacuum' clearouts.'
- ◆ 'In 1985 had operation to cover up hole in my eardrum.'
- ◆ 'When I was a teenager, I had my tonsils and adenoids out to try and improve my hearing - it didn't work.'
- ◆ 'Had tonsils out because I couldn't talk very well (they later found out I was deaf).'
- ◆ 'Had cysts behind ears removed surgically whilst very young but root bits can be a bit troublesome at times letting out pus when I'm run down a bit it seems.'

Other problems relating to ears/hearing:

- ◆ 'I had secretory otitis media at the age of 11 which took about 6 months to clear, and then not again until I was 48 years old. I now have recurring bouts of glue ear about every 4 months during which I experience mild conductive hearing loss.'
- ◆ 'Vertigo brought on as a result of several ear infections during late teens/early twenties. Consequent balance problems. Ears frequently completely wax up.'
- ◆ 'Suffer from wax build-up which is caused by a narrow tube to the ear and have poor hearing during the build-up. Have to keep having the wax removed. Had adenoids taken out aged 5.'
- ◆ 'Narrow ear channels (according to doctor).'
- ◆ 'I have a great deal of congestion in my head from time to time. My mouth is coated with catarrh. It seems connected.'
- ◆ 'I get very painful ears at times. This is sometimes associated with head and neck pains, and sometimes due to loud noises. The pain can be deep inside my ears and also round about them, both the bones and the soft tissues. The bones do grate on each other in this area. I sometimes need to massage and/or manipulate my ears in order to relieve pain and/or hear better again. I seem to be very oversensitive to noise and vibrations generally - 'hyperacusis'? I have to use ear-muffs in some situations.'
- ◆ 'My hearing is too sensitive. I have a problem with lots of noise.'

Comments relating to diagnosed hearing loss:

- ◆ 'Although recent hearing tests (at my request) show very little change, I feel distinction of sound (rather than volume) seems to be worse.'
- ◆ 'Hearing loss more likely to have been caused by noise induction.'
- ◆ 'Main weakness at ~4000Hz.'
- ◆ 'Even though I wear a hearing aid, I depend a lot on lip reading.'
- ◆ 'Had hearing aid aged 4 and hearing problems throughout childhood but have improved over the years.'

Comments relating to undiagnosed hearing loss/difficulties:

- ◆ 'Never been tested but feel over years that there is a minor loss.'
- ◆ 'I am to attend a neuro-otology clinic for the first time next month.'
- ◆ 'I have not been formally diagnosed as having a hearing problem - I am awaiting a appointment with an ENT specialist in four months' time.'
- ◆ 'Hearing on low side of normal.'
- ◆ 'My hearing is starting to get poor on the phone.'
- ◆ 'Hearing difficulties seem to be more acute when there is background noise i.e. in a bar/club. Also difficulties occur in large rooms such as classrooms where slight echoes occur.'
- ◆ 'If someone is speaking and I am watching TV, I have to key in so to speak with left ear and then miss what is said on TV.'

BONES AND JOINTS (n=79)

Percentage of sample first diagnosed by a rheumatologist/orthopaedic specialist 0%

As a child:

Hypermobile joints	47%
Unusually prominent joints	25%
Club foot (<i>talipes equinovarus</i>)	4%
(of whom 1% had corrective surgery)	
'Knock knees' (<i>genu valgum</i>)	27%

As an adult:

Hypermobile joints	35%
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Joint pain:

Have joint pain	81%
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Where in the body:

Toes	20%
Ankles	28%
Knees	83%
Hips	55%
Fingers	48%
Wrists	42%
Elbows	28%
Shoulders	45%
Jaw	11%
Neck	44%
Upper back	30%
Lower back	64%
Other:	14%

(specified as:

Ribcage/chest

Feet

Head/cranial and facial bones

Shins

Thumbs)

Age of onset of joint pain:

Early childhood (10 and under)	16%
Puberty (11-18)	26%
Adult (19-35)	40%
Mid-life (36-60)	16%
Aged 60+	2%
Not sure	5%

Frequency of joint pain:

All the time	22%
Varies (<i>according to level of activity, medication etc.</i>)	73%
Hardly ever	2%

[N.B. In addition to the above, eight respondents indicated that their joints had been particularly painful during pregnancy.]

Factors considered by respondents to affect joint pain:

a) Weather 69%

Respondents consistently mentioned cold, damp and extremes of temperature (including heat) as aggravating factors. Typical comments include:

- ◆ 'Cold, wind and damp all affect me badly. Extreme cold makes my bones start grating. Warm weather helps. Hot, dry weather helps a lot!'
- ◆ 'Heat makes my joints swell. Damp makes movement difficult.'
- ◆ 'If I'm out walking and my knees get damp - aaagh!'
- ◆ 'My knees seem to be more painful in the cold winter months.'
- ◆ 'Any change in weather affects my joints, especially inconsistencies i.e. hot one day, cold the next.'
- ◆ 'Extremes of heat/cold seem to stress difficulties making it much harder to cope.'
- ◆ 'Damp, humidity, rain... I can tell in the morning if it's going to rain that day.'
- ◆ 'Body likes low humidity and warmth.'

b) Time of day 64%

Respondents consistently indicated that early morning and late afternoon/evenings were the worst times of day for joint pain, particularly after work or an active day, although a few experienced pain night and day. Typical comments include:

- ◆ 'Always worst first thing in the morning.'
- ◆ 'Getting out of bed first thing in the morning my joints are stiff and painful.'
- ◆ 'Generally worse in the morning, easing up later.'
- ◆ 'When I'm tired or when I've been working they tend to hurt.'
- ◆ 'Early morning pains worst but shoulder and back pains remain all day.'
- ◆ 'Usually more mobile in the mornings, but problems can vary. Sometimes stiff, sometimes very hypermobile and uncoordinated. Pain levels vary considerably.'
- ◆ 'If painful, usually becomes progressively worse during the course of the day.'
- ◆ 'Second half of day is worse.'
- ◆ 'Night time, especially lying in bed.'
- ◆ 'All day and night.'

c) Exercise 83%

Respondents tended to fall into two groups - those who felt that gentle exercise was important for maintaining mobility, and those for whom any form of exercise or non-strenuous physical activity/movement caused pain and stiffness. Typical comments include:

- ◆ 'Exercise can help or aggravate depending on circumstances (e.g. initial degree of pain, stiffness, hypermobility etc.)'
- ◆ 'Regular exercise helps. Excessive exercise makes pain worse.'
- ◆ 'Gentle swimming excellent for increasing strength of muscles. Better muscles, less joint duty?'
- ◆ 'Need regular exercise, otherwise joints are more painful.'
- ◆ 'Helps to do gentle exercise.'

- ◆ 'It hurts while I'm exercising but doesn't get worse as a result - keeps me mobile.'
- ◆ 'During and after exercise joints stiff and painful.'
- ◆ 'Exercise increases hip and back pain mostly.'
- ◆ 'Exercise makes it worse and hurts spine and hip.'
- ◆ 'Intensive swimming training (around age 20) made knee and leg pains a lot worse.'
- ◆ 'Exercise like running or badminton tends to 'inflamm' (it feels like it) joints around the ankle and tendon. I do tend to walk slightly on the outside of my feet which might not help when exercising.'
- ◆ 'After extended physical activity, and a period of rest (even with stretching the muscles) I can put no weight on my knees.'
- ◆ 'When I play bowls, the pains increase in my back and shoulder.'
- ◆ 'Walking and housework make things worse. No longer able to 'keep fit'!
- ◆ 'Walking too fast, walking uphill and up stairs and steps...'
- ◆ 'Pushing pushchair, lifting anything heavy (e.g. 2 year-old daughter) affects my wrists and elbows. Hillwalking, especially going downhill, is a problem...'
- ◆ 'I have to walk quite a distance to collect my children from school - this can sometimes be very painful.'
- ◆ 'Cannot bend, stretch or do repetitive movements. Find it difficult to hold a newspaper or book, fasten buttons, lift saucepans... Cannot stand, sit or walk for long.'
- ◆ 'I can no longer kneel down easily and it is difficult to get up from my knees without support. It is also difficult to get up and down high steps e.g. on and off buses.'
- ◆ 'For a person my age (21 years) I have low stamina. I always had difficulty in PE at school. When I'm having a bad day, I sometimes cannot physically move.'
- ◆ 'My joints hurt when sitting, standing, or just moving around.'
- ◆ 'Joints become sore when I try and do any physical activity.'
- ◆ 'Due to mechanically unstable joints, all activity pushes chronic pain into acute phase; have fainted as a consequence. Haemorrhage after inadvertent damage to joints.'

d) Rest

62%

A few respondents found rest helpful, and several felt they needed more rest than their lives allowed. However, the majority indicated that resting, sitting or lying still for any length of time caused pain and stiffness, particularly on moving again. Typical comments include:

- ◆ 'If I rest for too long, I stiffen up and my joints lock, causing pain.'
- ◆ 'After I have sat down, it is very painful when I get up again.'
- ◆ 'If I sit for a long time, I have pain.'
- ◆ 'Knees become stiff if I sit for too long.'
- ◆ 'Too much inactivity causes more pain'.
- ◆ 'Movement after inactivity causes above average pains for a bit.'
- ◆ 'Even when resting, back, neck and legs are very painful.'
- ◆ 'Overnight stiffness in back. If I rest for a while, walking is then difficult.'
- ◆ 'After sleep, my back takes a little time to warm up.'

- ◆ 'Too much rest in one position makes joints worse. Wake approximately every hour at night and turn into another position. Sometimes go back to sleep, sometimes not.'
- ◆ 'Pain on turning during the night, pain and stiffness on waking, pain on sitting in one position for a few minutes.'
- ◆ 'Improves pain, especially raising legs, but can't sit still for too long. Can't lie on side (right or left) for a long period.'
- ◆ 'Rest with my legs up for a couple of hours a day.'
- ◆ 'I seem to need an awful lot of rest.'
- ◆ 'I need to rest when in acute pain and to take frequent short breaks when involved in physical activities. I need to rest for a long spell after moving around for a while.'
- ◆ 'Enforced bed rest to aid recovery and correct spinal posture. Rest joints before/after activity. Energy reserves needed to deal with acute pain so try to avoid getting overtired.'
- ◆ 'Rest helps, but hard with too young children and a blind mother.'

e) Medication

59%

No medication was indicated by any respondent to have significantly alleviated long-term joint pain. Some respondents gained temporary relief from paracetamol, ibuprofen and other over-the-counter painkillers or anti-inflammatory tablets/gels. Dosage varied considerably between respondents.

The most frequently named prescribed drugs, felt to be effective by some respondents but not by others, included naproxen, co-dydramol, meloxicam, diclofenac and arthrotec. Other medication mentioned once only included (in alphabetical order): acupan, aspirin, brufen, co-codamol, co-proxamol, cortisone injections, difflam cream, indomethacin, ketoprofen, losine, morphine sulphate, solpadol, tylex, voltaran. Arnica and rhus tox were used by one respondent.

Several respondents commented on adverse reactions to anti-inflammatory and/or prescribed drugs which had been severe enough to cause them to stop taking the drug. Side effects listed ranged from 'Tiredness', 'Drowsiness', 'Stomach cramps' and 'Nausea/indigestion' to 'Depression and moodiness' and 'Bleeding'.

In trying to describe drug effectiveness, many respondents indicated inadequate or unreliable pain relief. Typical comments included:

- ◆ 'The tablets lessen the pain and substitute it with a dull ache.'
- ◆ 'Doesn't take the pain away - just dulls it slightly.'
- ◆ 'Sometimes they help, sometimes not.'
- ◆ 'Have tried various prescription and over-the-counter drugs (can't remember names) but none seem to have a noticeable effect.'

f) Physiotherapy

39%

A few respondents had found that physiotherapy/hydrotherapy had offered short-term relief. However, the majority were less fortunate, with several indicating that physiotherapy had led to increased pain. Typical comments include:

- ◆ 'The hydrotherapy pool helps.'
- ◆ 'I had special physio which helped.'

- ◆ 'Hydro pool and gentle exercise can help for a short time until I get a bad flare-up.'
- ◆ 'Has helped initially, but after a month I am back to square one.'
- ◆ 'Had physiotherapy several times but only temporary easing of pain.'
- ◆ 'Had a course of physiotherapy on my left knee which is the most affected. It did give relief but only lasted a couple of months beyond the physio.'
- ◆ 'Did not find it particularly helpful.'
- ◆ 'Had physiotherapy some years ago but had to give it up - joints just too painful. Bad for weeks after.'
- ◆ 'I had a course of physiotherapy which caused me more pain.'
- ◆ 'Made joints worse.'
- ◆ 'Not any good - makes everything worse.'
- ◆ 'Made my hips and back much more sore.'
- ◆ 'Inappropriate hydrotherapy unfortunately resulted in me having to attend casualty. Ultrasound and laser moderately effective.'
- ◆ 'They gave up on me saying they can't help me any more.'

g) Other

31%

Respondents were invited to list other factors which affected their joint pain. Aggravating factors listed included the following:

- ◆ 'Any form of stress.'
- ◆ 'Symptoms worse in the week before a period and the first two days of it. Joints swell (particularly ankles and knees) and are painful.'
- ◆ 'Monthly period. One week before I get pain in hip, pelvis, back and knee and have difficulty getting around. Have a very bad headache, very tired, feel sick.'
- ◆ 'Tiredness/stress/illness can trigger joint pain. If I get a common cold my joints react.'
- ◆ 'Cold drafts.'

Alleviating factors listed included the following:

- ◆ 'Hot baths are great!'
- ◆ 'Massage helps.'
- ◆ 'Osteopathy. I have found this very helpful. My osteopath also practises acupuncture which can be helpful too. I prefer osteopathy perhaps because of its tactility.'
- ◆ 'Acupuncture only leads to temporary easing. Aromatherapy massage more effective but symptoms return.'
- ◆ 'Chiropractic treatment helped between 1981-6 in relieving pain and learning about my disability but may have contributed to my present hypermobility problems long-term.'
- ◆ 'If I lie flat out, ankles over the end of the bed, arms semi-raised, take a solpadol tablet and sleep for a couple of hours, I feel fine again.'
- ◆ 'Steroid injection into left knee. Good for about three months.'
- ◆ 'Supplied with supportive insoles for fallen arches. I only use them with slippers (have only had them for a month so far). Difficult to fit in normal shoes. Seem to be beneficial.'
- ◆ 'Controlled by diet. Fish oil and other health products also help.'
- ◆ 'Dr. Wallach minerals seem effective. Have reduced pain over a two-month period.'
- ◆ 'I try to walk every day and walk as a hobby.'

Other joint problems:

Joint stiffness		66%
Joints which 'lock'		32%
Joint dislocation		30%
	(of which:	
	hip	37%
	shoulder	47%
	knee	32%
	elbow	26%
	fingers	10%
	thumb	10%
	wrist	5%
	jaw	5%
	other: subluxations of vertebrae and sacro-iliac joints	5%)

Balance problems 35%

Typical comments include:

- ◆ 'Not very secure unless both my feet are on even ground.'
- ◆ 'Especially when tired - appear to be drunk when walking.'
- ◆ 'Very unstable when standing.'
- ◆ 'My balance is poor when I walk, stand still or kneel.'
- ◆ 'Getting up - sitting to standing and vice versa. Walking when tired.'
- ◆ 'Impossible to stand or balance only on one leg. Difficulty standing with or without crutches for long without losing balance even when legs are level.'
- ◆ 'Problems standing on one leg. At 14 months, walked with arms out all the time for balance and kept falling over until aged 4/5 when fitted with special boot.'
- ◆ 'One leg is shorter than the other. When the carer gets me dressed, I go off balance sometimes.'
- ◆ 'Have to hold onto things.'
- ◆ 'Sometimes lose balance.'
- ◆ 'Have always had a poor sense of balance.'
- ◆ 'May stumble or tilt but have never fallen over. Almost like I am drunk.'
- ◆ 'I have recurring bouts of vertigo for which I am on medication.'
- ◆ 'Only mild - related I think to migraines and fainting/dizzy spells.'
- ◆ 'Prior to becoming wheelchair dependent I used crutches and was unstable. Have fainted on occasion.'

Joint problems affecting mobility/everyday life 72%

(of whom affected in the following ways:

Length of time able to stand	52%
Distance able to walk	56%
Ability to climb steps or stairs	55%
Ability to look after self	20%
Job or choice of career	36%
Choice of sport/leisure activity	61%
Other:	

- ◆ 'Getting in and out of bath.'
- ◆ 'Sex.'
- ◆ 'Housework.'
- ◆ 'How I pace my week.'

- ◆ 'Interrupted sleep.'
- ◆ 'Extreme difficulty travelling.'
- ◆ 'Cannot go out on my own.'
- ◆ 'Have had to be medically retired.'

Joint problems worsened with age: 68%

Dependent on mobility and other aids 20%

(e.g. wheelchair 9%

sticks/elbow crutches 11%

neck/back brace 3%

knee supports 9%

wrist supports 3%

other aids to independent living 6%)

X-rays in connection with Stickler syndrome: 43%

(of which: full skeletal x-ray 35%

knee(s) 35%

hip(s) 29%

spine 26%

hand(s) 12%

wrist(s) 9%

face/jaw/skull 9%

elbow(s) 6%

other: shoulder

chest

ankles)

Typical x-ray findings include:

- ◆ 'Widened bone ends.'
- ◆ 'Dumb-bell shaped bones.'
- ◆ 'Bone degeneration.'
- ◆ 'Slight malformation of some of my lower vertebrae.'
- ◆ 'Tibia and fibula of left leg bent outwards. Had operation to straighten the leg.'
- ◆ 'Enlarged joints and odd fragments of bone.'
- ◆ 'Small fragments of splintering bone in the joints.'
- ◆ 'Worn ball joints.'
- ◆ 'Wear and tear of joints.'
- ◆ 'Hyper extensible joints.'
- ◆ 'Severe osteoarthritis of knees, lower spine and neck. Moderate OA in hips and ankles.'
- ◆ 'Early stages of osteoarthritis in my spine and myelitis.'
- ◆ 'The cup on the left elbow had fractured in several places. Osteoarthritis was present.'
- ◆ 'Osteochondritis of the hip and spine.'
- ◆ 'Curve at bottom of spine.'
- ◆ 'Scoliosis.'
- ◆ 'I had several x-rays around the age of 12-13 years for my knees locking although at this time I had not been diagnosed with Stickler syndrome and nobody could tell me what was wrong.'

Biopsy of the synovia 3%
 (When asked what the biopsy showed, one respondent replied:
 ♦ 'Membrane was breaking up.')

Joint replacement 9%
 (of whom: both hips (twice) 29%
 both hips (once only) 29%
 single hip (three times) 14%
 single hip (once only) 29%
 knee 14%)

Age at joint replacement:
 Aged 40-49 57%
 (of whom two had both hips replaced again at age unspecified)
 Aged 50-59 14%
 Aged 60-69 14%

[N.B. One respondent not included in the above figures had the same hip replaced three times - at ages 14, 28 and 44 - and a knee replacement aged 42. Another respondent commented:

- ♦ 'When I had a hip replacement, the bone kept breaking. Found I had soft bone so it was difficult to do and I still have problems.']

Other surgery for joint/musculo-skeletal problems: 14%

Respondents reported the following additional surgery:

- ♦ 'Surgery aged 16 for osteochondritis dissecans (left knee).'
- ♦ 'Hammer toe - straightened and pinned.'
- ♦ 'Two operations aged 1 to try and correct hammer toes but they were not successful.'
- ♦ 'Bone shaved on both feet.'
- ♦ 'Prominent heel - excess bone had to be removed.'
- ♦ 'Osteotomy to straighten left leg. Operation to release trapped nerve in left arm.'
- ♦ 'Surgery to straighten legs.'
- ♦ 'Fusion of bones in finger of right hand.'
- ♦ 'Elbow surgery (right) twice aged 10-11 to tighten ligaments after repeated dislocations. Faulty surgery has led to some lack of movement now and intermittent pain.'
- ♦ 'Surgery to reinforce ligament on right knee to attempt to eradicate movement of knee joint.'
- ♦ 'Keyhole surgery on left knee for torn cartilage and to clean out knee joint which has arthritis.'
- ♦ 'Had curettage (drilling into head of femur) on right hip.'

Scoliosis 18%
 Pectus carinatum (*pigeon chest*) 9%
 Pectus excavatum (*funnel chest*) 4%
 Fibromyalgia 9%

Other comments and problems with bones and joints:

- ◆ 'Osteoporosis of the spine.'
- ◆ 'Spondylo-epiphyseal dysplasia.'
- ◆ 'Born with a hump back (lordosis?)'
- ◆ 'Kyphosis; fibrositis; spondylosis; spondylitis; osteophytes; bruising around joints after moderate use; Marfan-like appearance.'
- ◆ 'Spondylolisthesis of spine, spondylosis of neck, osteochondritis of knees, arthritis in joints.'
- ◆ 'I have problems with my feet which make finding shoes a problem. This has not been medically assessed but probably should be.'
- ◆ 'Large feet (size 11) for a female.'
- ◆ 'No feeling in toes/ankles.'
- ◆ 'Rheumatoid arthritis.'
- ◆ 'Sciatica in my right leg.'
- ◆ 'Knee joints are very 'dry' and make loud scratching noises when climbing hills or stairs.'
- ◆ 'Joints which crack and click.'
- ◆ 'My neck frequently cracks so loudly that others can hear it.'
- ◆ 'Severe fatigue when my joints are particularly inflamed which affects my quality of life.'
- ◆ 'Had a tumour removed (periosteal osteosarcoma) from bone in left shoulder aged 17 years.'
- ◆ 'Painful joints are intermittent, although some things (kneeling) are always painful. 'Playing up' phases seem to have no set cause (apart from cold) and are getting progressively worse - hips were never a problem until the last one or two years.'