

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

UK CHILD DATA

STICKLER SYNDROME QUESTIONNAIRE SURVEY

November 1999

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UK CHILD DATA

- ◆ Data was gathered from 74 children (aged 16 and under) living in the United Kingdom.
- ◆ 38 (51%) respondents were male; 36 (49%) were female.

GENERAL

Diagnosed as having Stickler syndrome by a medical professional

Yes	97%
No	3%

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

Age at diagnosis

At birth/neonatally	24%
Neonate to 5 years	33%
Aged 6-10	29%
Aged 11-16	7%

Average age of respondents	9.12 years
Average age at diagnosis	4.2 years

By whom first diagnosed

Ophthalmologist	42%
Geneticist	28%
Cleft surgeon	7%
Paediatrician	12%
Rheumatologist	3%

Diagnosed by professionals working together (e.g. ophthalmologist, geneticist and cleft surgeon, ophthalmologist and rheumatologist, paediatrician and geneticist etc.)
6%

Other:
'Mother/dentist.'
'Diagnosed when brother born.'

Diagnosis by ophthalmologist at first retinal detachment

80% of UK children who had experienced one or more retinal detachment had been diagnosed by an ophthalmologist. For 67% of these children, the age at diagnosis and age of first retinal detachment were the same, leading us to conclude that the initial detachment had provided the basis for a diagnosis. Two children were diagnosed by an ophthalmologist *before* their first retinal detachment.

Relatives with Stickler syndrome

70% reported relatives (living or deceased) with Stickler syndrome.

EYES (n=74)

Percentage of sample first diagnosed by an ophthalmologist 42%

Current vision:

Myopic	77%
Has vision in both eyes	74%
Does not have vision in both eyes	26%

Registration:

Registered as partially sighted	18%
Registered as blind	4%

Retinal detachment:

Have experienced retinal detachment	20%
Number of detachments per child:	
1 detachment	13%
2 detachments	20%
3 detachments	40%
4 detachments	7%
5 detachments	13%
6 or more detachments	7%
Average age of first detachment	8.1 years
Vitrectomy	67%
(of whom 50% 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 13%
- ◆ Stickler syndrome diagnosed before first retinal detachment; but did not seek medical help immediately 20%
- ◆ Stickler syndrome undiagnosed at first retinal detachment, but sought medical help immediately 33%
- ◆ Did not recognise symptoms of retinal detachment nor seek medical help immediately 33%
- ◆ Would now recognise the symptoms and seek medical help immediately 80%

Cryotherapy and laser treatment:

Cryotherapy treatment to the retina	9%
(of whom: cryotherapy as part of surgery for retinal detachment	0%
cryotherapy as a preventive measure	86%
cryo. as part of surgery AND as a preventive measure	14%)
Laser treatment to the retina	19%
(of whom: laser as part of surgery for retinal detachment	21%
laser as a preventive measure	57%
laser for glaucoma	0%
laser as part of surgery AND as a preventive measure	14%)
Cryotherapy AND laser treatment	7%

Glaucoma:

Have glaucoma 8%
(of whom 83% have glaucoma in both eyes)

Glaucoma treated with:

Eye drops	50%
Tablets	17%
Surgery	50%

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

Cataract:

Have (or have had) a cataract 24%
(of whom: bilateral cataracts 61%

both cataracts removed	22%
one cataract removed	28%
no cataract removed	50%

Lens implant following cataract surgery 22%

Hospitals currently attended for eye care/treatment:

Addenbrooke's NHS Trust, Cambridge	12%
Manchester Royal Eye Hospital, Manchester	9.5%
Moorfields Eye Hospital, London	8%
Sunderland Eye Infirmary, Sunderland	8%
Royal Victoria Hospital, Belfast	3%
Birmingham and Midland Eye Centre, City Hospital, Birmingham	3%
Royal Surrey County Hospital, Guildford	3%
Victoria Hospital, Kirkcaldy	3%
Lincoln County Hospital, Lincoln	3%
Great Ormond Street Hospital, London	3%

Hospitals cited once only:

England Royal Liverpool University Hospital, Liverpool
Arrow Park Hospital, Wirral
Royal Victoria Infirmary, Newcastle upon Tyne
Bradford Royal Infirmary
Birmingham Children's Hospital
Wolverhampton and Midland Counties Eye Infirmary
North Staffordshire Hospital, Stoke on Trent
Children's Development Centre, City Hospital, Nottingham
West Norwich Hospital, Norwich
Oxford Eye Hospital, Oxford
Royal Berkshire Hospital, Reading
Worcester Royal Infirmary
Hillingdon Hospital, Middlesex
West Middlesex University Hospital, Middlesex
Royal London Hospital, London
Western Eye Hospital, London
Guy's Hospital, London
Frimley Park Hospital, Surrey
Longquest Hospital, Hastings

Wales Singleton Hospital, Swansea

Scotland Royal Aberdeen Children's Hospital, Aberdeen
Yorkhill Hospital, Glasgow

Eire The Eye Clinic, Dublin

Four respondents gave 'Optician' as the source of their child's current eye care and treatment.

Other comments and eye problems:

Treatment-related comments:

- ◆ 'Oil used to hold retinal detachment in place while healing - oil removed after 8 weeks. Some oil droplets remain inside eye - recently caused some irritation to the eye.'
- ◆ 'Was myopic - now needs bifocal glasses for slight long sight as no natural lenses in his eyes (right eye has implant lens, left eye no lens).'
- ◆ 'Cryotherapy seemed to correct almost completely the albeit mild myopia of -3/4.'

Other problems identified by respondents:

- ◆ 'The question of cataract surgery came up at my son's appointment yesterday because it is impairing his vision considerably. We were told they will not operate until absolutely necessary because the risk to his sight is so great and they want him to see for as long as possible. In other words, the hospital is of the opinion that my son will be totally blind after a cataract operation because he has already had a retinal detachment and the band and the oil will not hold up to further surgery. Therefore, the outlook for my son is not very good at all.'
- ◆ 'My child's loss of sight in the right eye is in part due to band keratopathy - cornea no longer transparent.'
- ◆ 'Squint.'
- ◆ 'Had operation for squint at age of 5.'
- ◆ 'Squint corrected at age 2.'
- ◆ 'Recurrent squints. Surgery for these diagnosed eye deterioration.'
- ◆ '3 operations to correct squints in both eyes.'
- ◆ 'My daughter did have a slight turn (squint) in her right eye when she was very young but regular attendance at the local Eye Clinic and wearing patches rectified the problem. If she is very tired, sometimes that eye becomes turned again.'
- ◆ 'My son has nystagmus and has recently had squint correction surgery.'
- ◆ 'Cataracts are congenital. Also has nystagmus.'
- ◆ 'My daughter has no central vision in right eye.'
- ◆ 'My daughter's eyes are different colours - one is blue, the other quite greeny-blue.'
- ◆ 'As a baby, had a small operation for a blocked tear duct.'

Social difficulties:

- ◆ 'My child has worn soft contact lenses since he was 10 months old (glasses at 5 months). To date the contact lenses have been very successful in two ways - they correct eyesight, and my son is not stigmatised by wearing glasses with thick lenses.'
- ◆ 'In my daughter's theory exams and in piano exams she needs the work to be in large print due to her myopia. Is becoming frustrated with this because it singles her out from the rest.'
- ◆ [*Comment in response to the question, "If your child had another retinal detachment now, would s/he seek help immediately?"*] 'She would find it hard to convince staff at school. Staff would think she wanted to miss a class.'

MOUTH AND FACE (n=74)

Percentage of sample first diagnosed by a cleft surgeon 7%

Cleft palate:

Born with a cleft palate 49%

Type of cleft:

Submucous 33%

Other 67%

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

Soft palate only 42%

Hard and soft palate 29%

Hard palate only 8%

In back half of mouth/posterior cleft 8%

Midline - Pierre Robin 4%

Don't know - clearly visible 4%

Don't know 4%

Other terms used by respondents in connection with their child's cleft included 'Horse-shoe', 'Full', and 'Bilateral complete'.

Age of diagnosis of cleft:

At birth 69%

Neonatally 22%

Age 3 years and above 8%

[N.B. 'Neonatal' indicates that diagnosis occurred between 1 and 14 days after the child's birth. The three children whose clefts were diagnosed after the age of 3 - one at age 4, one at age 8, and one at age 11 - all had submucous cleft palates.]

Age at which cleft repaired:

6 months and under 19%

7-12 months 47%

13-18 months 17%

19 months and over 8%

[N.B. Two respondents gave two ages for cleft repair - 6 and 14 months, and 1 and 4 years - but only the age of first repair has been included in the above figures.]

Of the cleft repairs on children aged 19 months and over, the actual ages of the children concerned were 2½, 3 (twice) and 8 years. The latter had a submucous cleft palate which was diagnosed and repaired when the child was 8 years old.

Cleft not repaired 8%
(of whom all had a submucous cleft palate).

Pierre Robin sequence:

Diagnosed as having Pierre Robin sequence	31%
Age at which diagnosis changed to Stickler syndrome:	
Aged 12 months and under	65%
Aged 13 months to 5 years	17%
Aged 6-10	9%
Aged 11-16	4%
Average age of change of diagnosis from Pierre Robin sequence to Stickler syndrome	2.1 years

Feeding, breathing and speech difficulties in childhood:

Had problems feeding as a baby	55%
(of whom 80% had been born with a cleft palate)	
Had problems breathing as a baby	39%
(of whom 75% had been born with a cleft palate)	
Had problems with speech as a child	54%
(of whom 67% had been born with a cleft palate)	
Had speech/language therapy as a child	51%
(of whom 74% had been born with a cleft palate).	

Oral/facial features:

High-arched palate	32%
Bifid uvula	9%
Flat midface with a broad and flat nasal bridge (<i>midfacial hypoplasia</i>)	65%
Prominent eyes	49%
Small vertical fold of skin from each eyelid down across the inner corner of the eye (<i>epicanthic folds</i>)	24%
Short bottom jaw (<i>micrognathia</i>)	54%
Teeth which do not bite together well (<i>malocclusion</i>)	45%

Surgery and/or treatment to the mouth or face:

Pharyngoplasty	12%
Nasal/midface bone or cartilage grafts	8%
Orthodontic treatment	31%
Surgery on the upper or lower jaw	1%

Other comments and problems with the mouth or face:

Other surgery/treatment:

- ◆ 'Had to have a tracheostomy at 2 weeks because he couldn't breathe because of small lower jaw and floppy epiglottis.'
- ◆ 'My son's jaw is coming forwards without surgery. He had surgery for reflux at 5 months old. He was in hospital from 6 hours old to 6 months old and in intensive care for 3 months.'
- ◆ 'No longer bifid uvula after repair. Pharyngoplasty may be necessary.'
- ◆ 'Bifid uvula stitched at 9 months.'
- ◆ 'She has to have many teeth out due to her gums and mouth being very small.'
- ◆ 'Removal of extra teeth - peculiarly formed.'
- ◆ 'Adenoids removed.'
- ◆ 'Multiple tooth extractions due to overcrowding. Will need midface surgery later.'
- ◆ 'Will have a pharyngoplasty soon.'
- ◆ 'Just about to have pharyngoplasty and orthodontic treatment.'

- ◆ 'Will probably need orthodontic treatment but this has not started yet.'
- ◆ 'Will need orthodontic treatment.'
- ◆ 'The doctors wanted to do nasal/midface surgery when she was a baby but only because she did not look as a normal baby does. Her bottom jaw is becoming shorter as she grows older. She has not had orthodontic treatment yet.'

Speech and respiratory problems:

- ◆ 'My daughter's breathing problems were due to the fact that she was unable to obtain correct saturation levels i.e. most people saturate at 97-100% but she saturated sometimes as low as 50% especially during sleeping times. This resulted in her drastically losing weight.'
- ◆ 'Nasal speech.'
- ◆ 'Speech problems until cleft repaired.'
- ◆ 'Speech therapy done automatically to ensure words were pronounced clearly (first language Welsh until school age.)'

Other oral/facial problems:

- ◆ 'Often has what seems to be sinusitis and nasal discharge.'
- ◆ 'Lower eyelids thicker than usual.'
- ◆ 'Delayed eruption of deciduous teeth and enamel hypoplasia. Large interdental spacings.'
- ◆ 'Missing teeth.'
- ◆ 'Teeth very weak.'
- ◆ 'Dental decay - 9 milk teeth removed just before age 3. Decay onset at 15 months. Was told he was tongue-tied at birth.'
- ◆ 'Has an underbite.'
- ◆ 'Late to talk - first words aged 2½. Orthodontist took x-rays of jaw - showed immature dental development.'
- ◆ 'Possible verbal dyspraxia.'
- ◆ 'Food and drink come down nose. Tendency to choke.'
- ◆ 'Eldest daughter and husband have a high-arched palate (neither are diagnosed).'
- ◆ 'Pierre Robin was not diagnosed but was considered.'
- ◆ 'Pierre Robin sequence not diagnosed but Robinow syndrome (sic) was. Diagnosis changed to Stickler syndrome at age 2.'

Improvement of appearance with age:

- ◆ 'Midfacial hypoplasia more pronounced when aged about 5 with hardly any bridge to her nose.'

HEARING (n=74)

Percentage of sample first diagnosed by an audiologist 0%

Ear infections:

Frequent ear infections 38%
Diagnosed with 'glue ear' (*otitis media*) 51%
(of whom 11% were treated by antibiotics only, 39% by the insertion of grommets only, and 45% by both antibiotics and grommets. 5% recorded no treatment.)

Hearing loss:

Hard of hearing 51%

Hearing loss in one ear only 11%
(of whom: mild 25%
moderate 75%)

Bilateral hearing loss 89%
(of whom: bilateral mild 50%
bilateral moderate 38%
bilateral severe 5%
mild/moderate 3%
moderate/severe 3%)

Nature of hearing loss:

Conductive only 24%
Sensorineural only 18%
Conductive *and* sensorineural 34%
Don't know 24%

Age when hearing loss first diagnosed:

Aged 0-3 months 11%
Aged 4-6 months 13%
Aged 7-12 months 26%
Aged 13 months to 5 years 34%
Aged 6-10 years 13%

Hearing loss worse now than when first diagnosed 29%

[*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 11%
In one ear only 25%
(of whom: mild 50%
moderate 50%)
In both ears 75%
(of whom: mild 100%)
In head 0%

When asked to describe the noises heard:

- ◆ 'High-pitched buzzing.'
- ◆ 'Buzzing noise.' (X2)
- ◆ 'Ringing.' (X2)
- ◆ 'Like rushing wind and popping.'
- ◆ 'Loud - like lightning.'
- ◆ 'Bleeping in both ears.'

Hearing aids:

Wear a hearing aid for diagnosed hearing loss 37%
(of whom: In one ear only 7%
Bilateral 93%)

Value of hearing aid(s) as perceived by the wearer:
Help(s) a lot 64%
Help(s), but only in certain situations 36%
Do(es) not help at all 0%

Other comments and problems with the ears and hearing:

Relating to surgery/treatment:

- ◆ 'Perforated eardrum required surgery aged 12.'
- ◆ 'Unsuccessful grommet operations resulted in perforated eardrums. Each specialist (e.g. plastic surgeon and ENT consultant) had different opinions about the value of grommets.'
- ◆ 'Hearing is now good following insertion of grommets 2 years ago. A severe infection last year has left a perforated eardrum.'
- ◆ 'As a young child, had frequent ear infections and glue ear - treated by antibiotics and grommets in both ears aged 5-6. Did have a severe ear infection aged 11 which developed into a perforation of the eardrum - cleared by antibiotics.'
- ◆ 'She has perforated eardrums which need surgery to close at approx. 13-14 years. Wears the aliceband type of hearing aids and these are causing the problems of infection at this present time.'
- ◆ 'Frequent ear infections subsided after cleft repair.'
- ◆ 'Used to have frequent ear infections and glue ear when younger. Treated with antibiotics and grommets.'
- ◆ 'My son has his 3rd lot of grommets in place. But in the winter he will still experience some hearing loss so I use hearing aids as well. The hearing aids were only given to shut me up as I was not going to be fobbed off with, "Let's wait and see"!'
- ◆ 'My son is on set of grommets no. 7.'
- ◆ 'Has slight glue ear. GP is not keen on antibiotics and only prescribes if the eardrum is red. She is encouraged to blow her nose frequently.'

Comments relating to diagnosed hearing loss:

- ◆ 'Some high-tone deafness diagnosed.'
- ◆ 'Wears transmitter in school - radio system CRM 200.'
- ◆ 'Use of radio aid at primary school was very successful.'
- ◆ 'Aided at school only - has a lot of useful hearing. Used to have frequent ear infections but not now.'
- ◆ 'My daughter says she does not need hearing aids and is always very reluctant to wear them.'
- ◆ 'Did have a hearing aid but did not find it helpful and manages well without.'

- ◆ 'High frequency conductive loss diagnosed at three months, but no longer hard of hearing.'
- ◆ 'Had hearing problems due to glue ear when caught colds. This has settled with age.'
- ◆ 'Had frequent ear infections as an infant. Fitted with hearing aids as a baby but were of no use. Grommets fitted twice. High frequency sounds are missed.'
- ◆ 'Glue ear was when very young.'
- ◆ 'It is difficult to answer these questions in a child so young. Without his aids he may not have learned to speak as well or as quickly as he has. On the other hand, I felt that his first aids were too loud, but this was only a gut feeling at the time because his level of hearing was felt to be severe at the time of diagnosis.'

Comments related to undiagnosed hearing loss/difficulties:

- ◆ 'I have taken my child to the doctor's to get his hearing checked before now but they thought he just needed them syringed. I have not pursued the matter.'
- ◆ 'No hearing problems yet, but kept an eye on by paediatrician annually.'
- ◆ 'Awaiting referral for glue ear.'

BONES AND JOINTS (n=74)

Percentage of sample first diagnosed by a rheumatologist/orthopaedic specialist 3%

Hypermobile joints	53%
Unusually prominent joints	31%
Club foot (<i>talipes equinovarus</i>)	8%
(of whom 66% had corrective surgery)	
'Knock knees' (<i>genu valgum</i>)	22%

Joint pain:

Have joint pain	46%
Where in the body:	
Toes	6%
Ankles	59%
Knees	82%
Hips	44%
Fingers	15%
Wrists	20%
Elbows	12%
Shoulders	12%
Jaw	3%
Neck	12%
Upper back	12%
Lower back	20%
Other:	12%
	(specified as:
	Legs
	Heels)

Age of onset of joint pain:

Aged 5 and under	41%
Aged 6-10	41%
Aged 11-16	15%

Frequency of joint pain:

All the time	6%
Varies (<i>according to level of activity, medication etc.</i>)	82%
Hardly ever	9%

Factors considered by respondents to affect joint pain:

a) Weather	50%
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Respondents consistently mentioned cold and damp as aggravating factors, with a significant minority also finding that warm/hot weather caused joint pain. Typical comments include:

- ◆ 'Particularly prone to joint pain on damp, misty days, especially knees, hips and back.'
- ◆ 'When it's cold, she suffers quite a lot. Hot baths and cream rubbed into the areas help as well as rest.'

- ◆ 'My son's joints hurt him when it is cold or wet. He has a tendency to just collapse on the floor because his joints get stiff.'
- ◆ 'Hot and cold weather. Most comfortable at about 68°F (20°C).'
- ◆ 'Warm weather causes my son's knees to hurt.'
- ◆ 'Heat is her main problem.'

b) Time of day

41%

Respondents indicated that joint pain tended to be worse first thing in the morning and in the evenings/at night when their child was tired. A few children experienced pain at other times too. Typical comments include:

- ◆ 'When her joints flare up, is stiff getting up in the morning and also in the evening.'
- ◆ 'Worse first thing in the morning, after sitting for a while and excessive activity.'
- ◆ 'Generally worse in the morning, easing up later.'
- ◆ 'Mornings and when she has been lying down for a while.'
- ◆ 'In morning and if he's has been doing a lot of walking.'
- ◆ 'Late afternoon after a day at school and after walking home.'
- ◆ 'Mainly in the evening, but if a lot of walking the night before, morning as well.'
- ◆ 'Depends on level of activity for the day but always worse at night/evening.'
- ◆ 'Joints hurt mostly at night time but also during the day.'
- ◆ 'Evening and night.'
- ◆ 'During the night.'
- ◆ 'It varies.'
- ◆ 'Any time of the day or night.'
- ◆ 'When he's tired he complains of his knees hurting. Sometimes it's bad and he ends up crying.'

c) Exercise

100%

It would appear that children suffer considerably for their determination to be active and keep up with their peers, as all respondents indicated increased pain after exercise, and many found that even non-strenuous activities such as walking led to pain. Typical comments include:

- ◆ 'Intensive exercise increases pain.'
- ◆ 'Pain following load-bearing exercise.'
- ◆ 'Any high-impact exercise causes pain.'
- ◆ 'More activity equals more pain/stiffness.'
- ◆ 'Tendon stretching can cause pain due to club feet. Has had bouts of tendonitis which require resting the tendon.'
- ◆ 'Back pain arises from him becoming 'twisted' where he cannot stand up straight. Suspect a sudden movement may cause it.'
- ◆ 'Knees click out of place.'
- ◆ 'When my daughter was born her feet were turned outwards. When she was two she walked on her ankle bones.'
- ◆ 'She walks with a limp. She has one leg 2" shorter than the other and she drags her longer leg.'
- ◆ 'When been walking, leg joints painful.'
- ◆ 'After and during PE. When walking too far.'
- ◆ 'Walking long distance.'

- ◆ 'Overwalking.'
- ◆ 'A lot of running, jumping etc.'
- ◆ 'Running, jumping, playing Twister!'
- ◆ 'When doing high jump.'
- ◆ 'Playing football, roller-blading, running, walking up or down hill.'
- ◆ 'Some sports are quite difficult - runs awkwardly, joints become stiff.'
- ◆ 'If he does PE at school he falls a lot.'
- ◆ 'Especially after games lessons in school where she has exercised more than normal - ankles, toes as well as her knees and hips pain her.'
- ◆ 'During exercise and writing.'
- ◆ 'Because there is so much that my son cannot do due to the threat of trauma to the head, when he does any exercise he gets sore joints.'
- ◆ 'She used to do ballet but had to give it up.'
- ◆ 'Hurts him to run but he still does.'
- ◆ 'All sports are painful but he insists on having a go.'

d) Rest 32%

Several respondents found rest helpful, although others indicated that resting, sitting or lying still for any length of time caused pain and stiffness, particularly on moving again. Typical comments include:

- ◆ 'Rest allows inflammation of the tendons to subside.'
- ◆ 'After school, he rests with his legs up.'
- ◆ 'She rests in the evening.'
- ◆ 'Still painful when resting.'
- ◆ 'If she sits in the same position for any length of time she is stiff.'
- ◆ 'When she sits down for a while her legs hurt. After a night's sleep she says her body aches and she needs to stretch and bend when she gets up.'
- ◆ 'Always stiff after resting, sleeping etc.'
- ◆ Wakes at night, occasionally crying.'

e) Medication 35%

The following medication was mentioned by respondents: ibuprofen (50%), brufen (17%), junofen (8%), paracetamol (17%). One respondent mentioned Movelat cream, another piroxicam. Method of administration (e.g. oral tablets/suspensions and rub-in creams), dosage and perceived effectiveness varied, with some children taking medication regularly, and others only when required. Comments included:

- ◆ 'Prescribed ibuprofen which she took for two weeks but stopped as it affected her stomach.'
- ◆ 'Says the medication makes him feel better although I see no difference.'
- ◆ 'Give medication when necessary. 50/50 chance of it working - depends on whether the pain is severe or not.'

f) Physiotherapy 38%

Only a few respondents whose child had had/was having physiotherapy indicated whether or not the child found it beneficial. Provision varied greatly, with some children having physiotherapy 3-4 times a week and others 'as and when needed.' Typical comments include:

- ◆ 'Had small amount of physio and occupational therapy aged approximately 2-3 years for lack of coordination.'
- ◆ 'Physio on leg muscles seems to have improved his stance and movement.'
- ◆ 'My daughter has physio every three months. The physio has also been into the school to explain as she gives her exercises which help strengthen the muscles.'
- ◆ 'Has physio 4 times a week at school to stretch and build up muscle around the knee and hip joints.'
- ◆ 'Has physio because his sister does and because of his tracheostomy.'
- ◆ 'Hydrotherapy pool once a week.'
- ◆ 'Hydro pool and gym.'
- ◆ 'Under a physio as and when needed.'
- ◆ 'A lot depends on the exercises.'
- ◆ 'Doing exercises that the physio has given him to do make it worse - tired, pain.'

g) Other 15%

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

- ◆ 'Tiredness, stress.'

Alleviating factors listed included the following:

- ◆ 'If she complains at school they let her off P.E.'
- ◆ 'Has keyboard to help fingers.'
- ◆ 'Orthotics in his shoes improve his movement and have reduced tenderness in his hips and heels. The orthotics are purpose-made for my child and we suspect that the pains in his heels and hips were a symptom of feet problems that the orthotics are addressing.'

Other joint problems:

Joint stiffness		36%
Joints which 'lock'		13%
Joint dislocation		8%
	(of which:	
	hip	17%
	shoulder	17%
	knee	33%
	elbow	33%
	fingers	17%
	thumb	17%)

Balance problems 20%

Typical comments include:

- ◆ 'Very small feet and calcium deposits in the basal ganglia (responsible for balance and coordination).'
- ◆ 'Physiotherapist says he has low muscle tone.'
- ◆ 'Uneven hips and lack of muscle around them.'
- ◆ 'Due to leg length discrepancies and muscle weakness - physio helping to address this and posture therapy.'
- ◆ 'One leg shorter and thinner - wears boot with height.'
- ◆ 'She cannot balance on one leg.'
- ◆ 'If she stands for a while.'

- ◆ 'Especially when getting up first thing in the morning or after sitting for a while.'
- ◆ 'When sore with hip pain.'
- ◆ 'Due to being very clumsy on account of talipes.'
- ◆ 'His balance was bad as a young child. He has gradually improved this by practice (e.g. he is now a competent roller-blader).'

Joint problems affecting mobility/everyday life 39%
(of whom affected in the following ways:

Length of time able to stand	41%
Distance able to walk	79%
Ability to climb steps or stairs	41%
Ability to look after self	31%
Job or choice of career	31%
Choice of sport/leisure activity	79%

Other:

- ◆ 'Problems with advanced piano technique.'

Joint problems worsened with age: 24%

Dependent on mobility and other aids 7%

(e.g. wheelchair (for outdoor use) 3%

knee supports 1%

wrist supports 1%

orthopaedic boots 1%

other:

- ◆ 'Sometimes a sling for her elbow.'
- ◆ 'Use pushchair if walking is more than she can manage.'

X-rays in connection with Stickler syndrome: 32%

(of which: full skeletal x-ray 8%

knee(s) 29%

hip(s) 42%

spine 12%

hand(s) 21%

wrist(s) 4%

face/jaw/skull 8%

elbow(s) 4%

other: legs 29%

arms 25%

ankles 4%

feet 4%)

Typical x-ray findings include:

- ◆ 'Hyperextensible joints.' (X2)
- ◆ 'Head of femur badly out of shape.'
- ◆ 'Knuckle joints in hands fused as tips of bones are square not rounded and the limbs are very long. Other results not yet known.'
- ◆ 'The bones are not rounded, they are jagged.'
- ◆ 'Bilateral Perthes disease; epiphyseal dysplasia at left knee; bilateral ankle collapse. Jaw shows immature dental development.'
- ◆ 'Epiphyseal dysplasia.'
- ◆ 'Dumb-bell shaped bones.'

- ◆ 'Dumb-bell flaring.'
- ◆ 'Enlarged joints - more examinations to come.'
- ◆ 'Dislocation of left hip.'
- ◆ 'Kyphosis.'
- ◆ 'Helped the geneticist to determine the actual diagnosis of Stickler syndrome.'
- ◆ 'Awaiting result for bone age check.'
- ◆ 'Don't know. Was never sent any correspondence. The hospital in Oxford asked to see my son and his father purely for research.'

Biopsy of the synovia 1%
 (When asked what the biopsy showed, the respondent replied:
 ◆ 'Can't remember - done when she was a baby.')

Joint replacement 0%

Other surgery for joint/musculo-skeletal problems: 3%
 Respondents reported the following additional surgery:
 ◆ 'Open reduction of left hip to correct congenital dislocation.'
 ◆ 'Right talipes correction aged 5 months; right shelf acetabularplasty aged 5; right valgus derotation femoralosteotomy aged 6, left aged 7; bilateral ankle fusions aged 8.'

Scoliosis 1%
 Pectus carinatum (*pigeon chest*) 1%
 Pectus excavatum (*funnel chest*) 1%
 Fibromyalgia 1%

Other comments and problems with bones and joints:

- ◆ 'Broken thigh bone (spiral fracture) at age 2. Twisted leg when stepping on a toy; seemed to break surprisingly easily for such a minor accident. Maybe 'over-twisted' due to bilateral club feet? (one worse than other - has only had corrective surgery on one foot.)
- ◆ 'Three broken collarbones and a broken leg.'
- ◆ 'Still trying to confirm if my child has arthritis of the chest bone. Doctor says 'yes', rheumatologist said 'yes' and then 'no' after another appointment.'
- ◆ 'Poor muscle tone.'
- ◆ 'May have had joint pain before aged 2 but unable to express this. Stopped walking at about 2½ years old.'
- ◆ 'Has difficulty running and rising from sitting particularly from the floor. School PE and sports lessons cause him problems. Less agile than children of his age.'
- ◆ 'My daughter is unable to bend the joint at the base of her fingers although she can bend the joints at the knuckles. This applies to both hands.'
- ◆ 'Stickler syndrome first diagnosed because cannot make a fist at all - hand joints fused.'
- ◆ 'She has one 'curly' toe on each foot i.e. 2nd toe in from the little toe on each foot. She also has inserts in her shoes to prevent fallen arches.'
- ◆ 'Feet joints cause problems - sometimes feels the toes are curling under and has difficulty and pain when trying to straighten them.'

- ◆ 'As a baby (2 months) heels and ankles were noted to turn outwards. Physiotherapy and strapping corrected this by age of 15 months. Walked at 16 months - no further problems.'
- ◆ 'One foot turns in a little.'
- ◆ 'Joints 'crunch' - noise reverberates around the room.'
- ◆ 'Joints very loose and click loudly e.g. when dressing/undressing, particularly shoulders.'
- ◆ 'Joint mobility improving due to Vitamin D supplements.'
- ◆ 'Woken in the night by pain in her back along the spine.'
- ◆ 'My daughter was born with hip obliquity (outgrown), camptodactylia (outgrown), asymmetrical head and chest (outgrown).'
- ◆ 'VERY long fingers, toes, hands. Pronounced endings of bone under knee. Very bony altogether! Has always had joint pains - thought everyone did.'