

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

**FINDINGS OF THE UK AND EUROPEAN  
STICKLER SYNDROME QUESTIONNAIRE SURVEY**

## **SUMMARY**

**November 1999**

# **FINDINGS OF THE UK AND EUROPEAN STICKLER SYNDROME QUESTIONNAIRE SURVEY**

Stickler syndrome is a progressive genetic condition originally called 'Hereditary Progressive Arthro-ophthalmopathy' and has been estimated to affect up to 1 in 5,000 people. There are wide variations in the clinical manifestations of this disorder, even within a family group, and it is still greatly under-diagnosed.

In order to further define the expressivity of this syndrome, a confidential medical questionnaire study was undertaken. This was the first and largest questionnaire study on Stickler syndrome and involved members of support organisations in the UK (Stickler Syndrome Support Group) as well as in the USA (Stickler Involved People), Australia, The Netherlands and Canada.

## **Methodology**

A 19-page questionnaire was designed by Dr Gunnar B Stickler, Wendy Hughes (SSSG) and Pat Houchin (SIP) with additional input from the group in The Netherlands. The questions were reviewed by doctors from the UK specialising in ophthalmology, rheumatology, orthopaedics, audiology and oro-facial disorders.

The questionnaire had a section on General Information followed by more detailed questions on Eyes, Mouth and Face, Hearing and Bones and Joints. In total there were 60 major questions on the condition with numerous subsets. The study should be considered a subjective study (and not objective) as no work has been undertaken with a matched sample from the general population.

Of the 216 people associated with the SSSG who were asked to complete the questionnaire, 153 were completed and returned in respect of 79 adults and 74 children. The data for the European study is for 4 adults from The Netherlands and Germany. The high rate of participation was extremely encouraging and a collective thank you to all those who spent a lot of time and effort in completing and returning the questionnaires.

## **Conclusions**

The study has reconfirmed that early and proper diagnosis of the condition can significantly help the management of the condition and the medical outlook of Stickler patients. As Stickler syndrome is a complex, long-term disorder that can affect many of the body's systems, it is essential that there is a co-ordinated multidisciplinary approach to care provision.

The number of respondents who had received genetic counselling or testing in the UK was disappointingly low when compared with the experience reported by patients in the rest of the world study. It is becoming increasingly evident that proper counselling and testing are an integral part of the care management process and more effort needs to be made by all of us to help achieve this goal.

The age of diagnosis for adults at 32 years is relatively high and reflects the serious lack of awareness that existed within the medical profession. The level of awareness has increased, although there is still a long way to go, and some encouragement can be taken that the average age of diagnosis in the child survey is now 4.2 years. A priority of the SSSG awareness raising work will be to increase the focus on those medical specialities that deal with babies and younger children.

The serious sight and oro-facial problems associated with Stickler syndrome have again been confirmed by this study with 75 per cent of all respondents reporting myopia, 61 per cent of UK adults suffering retinal detachments and nearly half of all children respondents having a cleft palate.

Joint problems have also been shown to be a very significant issue, with joint pain being reported by over 80 per cent of all adult respondents. The level of pain reported varies significantly with the weather, time of day and level of exercise. In many instances these joint problems have resulted in Stickler patients being restricted in their daily mobility and having their choice of leisure activities and careers affected. Some respondents also noted an increase in pain after physiotherapy and this may indicate a need to assess the type of care management and possible surgical intervention that Stickler patients currently receive for their joint problems.

It was also interesting to note that nearly a quarter of all UK adult respondents suffered some form of tinnitus. This confirms previous anecdotal evidence and more understanding is needed about this distressing problem and how it can be managed most effectively.

## **Further Information**

The Stickler Syndrome Support Group has produced a comprehensive set of publications relating to the condition. If you need further information, please contact the group at the address below or you can visit our website: [www.stickler.org.uk](http://www.stickler.org.uk)

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## Summary of main clinical manifestations

The following table summarises the percentage of the respondents suffering from the clinical manifestations

	<u>UK Adult %</u>	<u>UK Child %</u>	<u>Euro Adult%</u>
<u>EYES</u>			
Myopia	76	77	100
Retinal detachment	61	20	50
Glaucoma	20	8	25
Cataracts (in one or both eyes)	64	24	74
<u>MOUTH AND FACE</u>			
Born with cleft	23	49	50
Initially diagnosed with PRS	10	31	50
<u>HEARING</u>			
Frequent ear infections	25	38	75
Hard of hearing	38	51	75
Tinnitus	23	11	50
<u>BONES AND JOINTS</u>			
Joint pain	81	46	100
<u>As a child:</u>			
Hypermobile joints	47	53	50
Unusually prominent joints	25	31	50
Club foot	4	8	25
Knock knees	27	22	50
<u>As an adult:</u>			
Hypermobile joints	35	-	50
<i>No. in sample</i>	79	74	4