by the signs, symptoms and the family history of the patient. This process is labour intensive as it requires a technician to search manually for the faulty gene. A team at Addenbrookes Hospital, Cambridge, is also investigating Stickler syndrome and has been conducting ophthalmic and genetic testing for a number of years.

Implications for nurses
- Nurses need to be aware of all the signs and symptoms associated with Stickler syndrome so that when problems group together this will not be attributed to coincidence. The boundaries of assessment need to be extended beyond each presenting complaint.
- Suspicions should be discussed in multidisciplinary teams because this is likely to lead to appropriate action being taken, such as further screening.
- Ophthalmic problems need to be treated urgently since any delay in treatment can result in irreversible damage. Stickler syndrome is the most commonly inherited cause of rhegmatogenous retinal detachment in children who have a high risk of retinal tear, which is commonly bilateral and a potential cause of blindness (Snead, 1996).
- Dental nurses need to be aware that a cleft palate may be part of a wider syndrome.
- School nurses have a compelling need to be up to date on all aspects of this condition because their client group may require urgent medical assistance.
- Stickler syndrome has no cure, so prompt diagnosis and prophylactic treatment is required to manage this complex syndrome. Once a diagnosis has been established the need for emotional and family support is of paramount importance.

Living with Stickler syndrome
The following is a personal account from someone who has recently been diagnosed with Stickler syndrome. The case study presents the personal and social issues connected with living with the condition and shows how the condition can go unnoticed for years. Nurses can help in this respect by taking a holistic approach to patient care and raising the possibility with colleagues that a patient may have the condition.

‘I am a 35-year-old psychiatric nurse, married with two children. A short time ago both my children and I were diagnosed with Stickler syndrome. My childhood was normal; I did well at school academically and I had no problems physically until, at the age of 12, the retina in my left eye spontaneously detached. As it was painless I did not seek help and, unfortunately, it was not until I sustained a shoulder injury at work and X-rays revealed that the ball of my shoulder joint was slightly irregular and further investigations showed that there were similar problems with some of my other joints. No reason was given for either the detached retina or the joint irregularities and no connection was made between these two incidences.

‘When my first child was born in 1998 it was noted at birth that he had a cleft of his soft palate. A geneticist who saw my son during his stay in hospital for cleft repair surgery at the age of eight months, felt that the cleft was an isolated problem. While reading up on cleft palate, however, I discovered a condition that links retinal detachment, cleft palate and joint problems. At this point, I was certain that both my son and I had this condition. However, no one else agreed, including my ophthalmologist, paediatrician, geneticist, orthodontist or my husband.

‘My daughter was born in 2000, also with a cleft soft palate. A consultant geneticist finally confirmed the diagnosis of Stickler syndrome in April 2002, but only after we pushed for an appointment to answer the mounting questions we wanted answered.

‘Once formally diagnosed I made contact with Wendy Hughes, president of the Stickler Syndrome Support Group, who has the syndrome herself. I had already read Hughes’ article (Hughes, 1995) and found that her support group provides excellent information and regular research updates.

‘I felt and continue to feel so many conflicting emotions. Fear for what the future holds for my children and worries about their eyesight, joints and problems that may arise at school. I feel angry that I have this condition and that I have passed it on to my children. However, I also feel relieved that we now know what the problem is and can seek help should any difficulties arise.

‘Would I have had my children if I knew about the syndrome? I don’t know, but I do know that I cannot imagine life without them. It gives me hope that I have lived with the condition for 35 years without it overshadowing my whole life. I have also been told that I do not “look” like someone who has Stickler syndrome. Certain facial characteristics can be present but are not always immediately obvious.

‘In our case, the condition seems to have started with me because we have no other family history. My chances of passing the condition on to my children are 50:50 for each child. This is also true for my children, unless genetic research takes a huge leap forward. The severity of the symptoms varies widely, even within the same family. My children are bright, articulate, energetic and a source of real joy and at some point in the future my husband and I will discuss this condition with them.

‘I decided to contribute to this article because I want to raise awareness of a relatively common, yet seriously underdiagnosed condition. Also, all the information I have read seems to be so frighteningly negative. However, research in this field continues and happiness while living with this syndrome is not an unobtainable goal.’

Further information about Stickler syndrome and an information pack may be obtained from:
Stickler Syndrome Support Group
PO Box 371
Walton on Thames
Surrey KT12 2YS
Tel: 01932 267635
www.stickler.org.uk

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