

## 2015 FAMILIES FOR FAMILIES DAY

### MEDICAL ADVISORY GROUP

#### **Question 1: What research is going on for adults aged over 30 with Noonan Syndrome, and where can they get help and advice?**

A study with a cohort of families started in 1985 and most of them are now young adults in their twenties. Hopefully the group can continue to be followed up so that we'll really know what's happening later in life.

There was also a collaborative effort across Europe to try to understand the natural history of the cardiac complications of Noonan Syndrome. Also clinically, there are a number of specialised centres for example cardiomyopathy expert centres in the UK, which have a lot of experience of looking after adults with Noonan Syndrome-related congenital heart disease.

The NHS Management Guidelines for Noonan Syndrome are also due for review this year and hopefully will include the lifetime management of the condition.

It was important to remember however that Noonan Syndrome was not progressive. There could be complications later in life but many people remain in good health with no further problems.

#### **Question 2: Where should a family go for support for a child with Noonan Syndrome and ADHD who hadn't had success going to CAMS and a psychologist whose only suggestion was medication such as Ritalin which their cardiologist said her daughter couldn't have with her heart defect.**

Obviously the specific answer depended on the specific cardiac manifestations that the child has. As a general rule it is true that medication like Ritalin can affect the heart – it can speed it up and it can sometimes cause abnormal heart rhythm. But one view is that it is a balance between how much the symptoms of ADHD are affecting the individual's quality of life balanced against the possible risk of an abnormal heart rhythm with the medication. What normally happens is that if medication is considered the right treatment for the ADHD, the treatment is started with very close cardiac monitoring - ECG and perhaps 24 hour tape - and in the majority of cases there aren't complications. Of course it depends on the specific condition and it's vital to know what the specific heart condition is but ADHD plus Noonan Syndrome should be a caution not a contra-indication when it came to medication.

In terms of support, the family were going down the best route in that they had a psychologist involved and CAMS but it was also important to get everyone to communicate well with each other which was a real key with behavioural issues. It isn't easy but is the best approach.

#### **Question 2: A parent who had one son who was PTN11 Noonan Syndrome and another son who had a clinical diagnosis. Last year he was retested for Noonan Syndrome and was found to have a chromosome sex duplication so she was considering whether to have her other son retested and wondered if it was possible for a Noonans child to carry another duplication.**

This is a possibility as just because you have a particular gene test result for Noonan Syndrome doesn't mean there wouldn't be other things. That can sound worrying but actually differences in genetic material are really common and if we look closely enough we've all got them. Larger differences that can have an effect are a bit less common but it would be wise to get it checked out.

**Question 3: The next question came from a parent whose baby had been born with extreme hydrops (increased fluid around the pregnancy). The obstetrician basically did a quick “Google” search as he had no guidelines for pregnancies for Noonan Syndrome and didn’t find anything that could go wrong but wondered if there were plans to write any guidelines?**

The problem is that hydrops occurs in a lot of different conditions so the problem for the obstetrician is what the underlying cause is. So they may look for a number of causes – infections for example – and Noonan Syndrome comes a long way further down the line and sometime doing the test can take some time in getting the results. Certainly a number of labs will offer the PTFN11 test for Noonan Syndrome if they think that’s a possible diagnosis in the womb. Hydrops is due to lymphatic problems so there have been a number of examples where Noonan Syndrome cases have presented with hydrops. Hopefully there a paper will be produced on this and particularly regarding prognosis and what’s best to do in the prenatal period.

**Question 4: What should parents do about a child in education who is borderline but not stated for special needs.**

Just because a child didn’t have a statement of educational needs or an Education & Health Care Needs assessment didn’t mean that their needs shouldn’t be met. Any child who is identified as having a special need or disability, is entitled under the Equalities Act to have education and support in school. So it’s about going back to the school and talking to them about it. It doesn’t matter if the child has a diagnosis or not, if they’ve identified some weak areas then the school should be addressing that through the SEN Support and should have a plan in place. They should be in that cycle of assess/plan/do/review and the parent should be involved in designing and reviewing that plan. The parent shouldn’t have to go on and on at a school but sometimes they do have to give them that nudge and keep going back in.

**Question 5: One child got very stressed about any challenges that the teacher sets and wasn’t getting the emotional support.**

One of the areas in the new Code of Practice was social, emotional and mental health issues and that emotional support is exactly what that covers. If she is having anxiety issues, school should be putting in strategies around that. If they don’t know what to do, they can get an Educational Psychologist to suggest strategies, but a lot of schools have buddying or mentoring groups that work in boosting self-esteem so there are things schools should be doing in addition to just “learning”.

**Question 6: A 5 year old has eating and speech problems, and suffers from reflux, has had a myo-rotation correction and his parents don’t believe he has any appetite whatsoever. Also the child’s speech and language specialists and dieticians aren’t familiar with Noonan Syndrome.**

From early on, it was recognised that feeding difficulties were part of Noonan Syndrome children and from that there is sometimes an impact on language development. There have been papers published on this and it may be that speech and language therapists might find that sort of information useful and NSA will try to make this more widely available.

It can be difficult getting a child to feed who might not be able to feed well or who has an aversion to feeding. It can become a bit of a family struggle and there can be a behavioural aspect developing from it. It’s useful to break down exactly what the problem is because it may be an immaturity in terms of swallowing, sometimes it can be tongue-thrusting - when you try to put food in, it’s pushed out. By analysing that, you can sometimes come to a programme of treatment with the therapist which will improve the feeding.

One parent found that giving a child with the tongue-thrusting issue, spicy and highly flavoured food eventually led to him starting to enjoy his food although it took time.

**Question 7: Is there research into children who are tube fed longer term e.g. at age 11 and is this physiological or behavioural.**

At the present time, research was at the stage of looking at patient surveys and considering anecdotal evidence as to what works. For some families the introduction of a blended diet through the peg tube seems to have been effective but there was no hard and fast rule and it was very difficult. It seemed certain that feeding and speech difficulties were closely related and to get anywhere with research, it would require psychologists and speech and language therapist all working closely together.

**Question 8: A girl with Noonan Syndrome was late in starting her periods and they were very painful. Was this unusual and what was the situation regarding fertility in females with Noonan Syndrome.**

Although there were no fertility issues with Noonan Syndrome, females did tend to start their periods later although they weren't necessarily heavy or painful.

**Question 9: A boy diagnosed at the age of 8, has a severely concave pectus or chest. Is this purely an aesthetic issue as the specialists say, should consideration of treatment wait till he reaches puberty or older so that it's his decision and is there a relevant connection with Noonans?**

The panel's understanding was that this was generally and usually a cosmetic issue rather than something that would cause a problem with the heart. It's very unusual for such a condition to compress the heart and cause a problem. Occasionally problems with the chest wall can cause problems with lung development but usually it's a cosmetic issue. The chest wall continues to develop as the child grows so it's often a good idea to wait until the child has stopped growing before making a decision.

**Question 10: Is a child's auditory neuropathy (hearing loss in which the outer hair cells within the cochlea are present and functional, but sound information is not faithfully transmitted to the auditory nerve and brain properly) as a result of having Noonan Syndrome?**

Some children with Noonan Syndrome have problems with hearing, most commonly "glue ear". It's possible that the child's condition was linked to Noonan Syndrome but more information was needed. There were genetic tests which should be able to tell if the condition was linked causally. People with Leopard Syndrome, closely related to NS, often suffered hearing problems.

**Question 11: A lot of NS children have had open heart surgery for pulmonary stenosis. Will future developments in technique mean a less invasive treatment?**

Less invasive techniques were available for more straightforward cases to open up a pulmonary valve but this "balloon" procedure didn't work as well when the valve was very thick or abnormally formed. In these cases surgeons often choose to go for open heart surgery as the result is likely to be better first time round. But in many individual cases of children with NS it was possible to do the less invasive balloon dilation technique and hopefully that these techniques will improve in the future.

**Question 12: A 4 year old boy has pains in his legs and arms and can wake up screaming in the night because of this but that the medical response was that they were just "growing pains". Is this the case?**

This was a fairly common issue, especially in adolescence when there could be a lot of muscle and joint pain. A small study in the past didn't show that there was any underlying problem which would cause long term damage. In most cases, the symptoms respond to normal analgesics such as Calpol or paracetamol or sometimes even a physical thing like a hot water bottle eases the local pain. It does tend to get better with time. It's a question which needs more research but at the moment the specific cause can't be pinpointed. There is a suggestion that many people with the condition are vitamin D deficient although in at least one case, treating this didn't seem to alleviate the problem.

**Question 13: Is it common to have an issue with a child regulating his body temperature – starting to go blue after 5 minutes swimming but so hot in bed at night he can hardly be touched?**

The issue with body temperature is seen in neurological conditions such as CFC and Costello and sweating seems to be quite common in Noonan Syndrome but it does seem to diminish with age.

**Question 14: If a child was tested in the past when they couldn't identify the gene, should she be retested and if so, if the child isn't seeing a paediatrician, should this be arranged through the GP or straight to the genetic centre?**

The number of genes identified keeps increasing so if tests were done some years ago it could be more specific results from a new test would be helpful. Paediatricians should be able to do the tests but in this case, the approach should be through her GP because that was the usual way it was organised in the Health Service and the GP can make a direct referral. To the regional genetic centres.

**Question 15: How open are doctors now to screening embryos for something like Noonans?**

This is very much a question of personal choice. The GP can talk through the options with the person. The normal test is 9 or 10 weeks into pregnancy but even though it can indicate the gene is there it can't always tell how severe the Noonans is going to be, and even in multi-generational cases, there may be differences between one generation and another. All this needs to be discussed sympathetically by the GP with the person in question.

**Question 16. A boy bruises very easily when he falls over and the bruises last a very long time. He's unfortunately got both pulmonary and aortic valve problems so he'll need an artificial valve when his heart deteriorates to a certain level and then he'll have to go on blood thinners. What considerations would there need to be regarding his bruising issues.**

There has been a fairly large study of clotting factors in children with Noonans which found that there were occasional issues with factor 8 clotting agent but usually these were not causing serious bleeding although they did cause bruising more easily. In rare cases of post-operative bleeding, it was relatively easy to correct that with fresh blood which did contain the clotting agent. If it did come to an operation for the child in question, the specific needs would be looked at carefully and there were options as regards blood thinners as new ones were being developed, although very few were licenced for use with children at the moment. There are artificial valves which don't require anti-coagulation but as in most cases, it very much depended on the individual case and there would be very detailed investigation before a decision was made. There had however been several cases where Warfarin had been used with children with Noonans.

**Question 16: A boy with Noonans hadn't put any weight on for 2 years although he is growing. The dietician had said to put him on a high calorie diet, basically to give him as much fat as possible can. Was this the best thing to do?**

Usually the first and most crucial question when a child's weight wasn't increasing, was whether the child was growing. Many young people with Noonan Syndrome were lightly built. There are many issues to consider with diet as a balanced diet is usually preferred and there is also the issue of the lymphatic function in coping with fatty foods which can be compromised in some people with Noonans.