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SCREENING FOR COELIAC DISEASE IN CHILDREN WITH DOWN SYNDROME

Dear Parents,

It is well established that children and adults with Down Syndrome have a higher risk of developing coeliac disease, compared to the general population. The risk is thought to be about 1 in 20 (5%), compared to 1 in 100 (1%) in the general population. Currently children with Down Syndrome are routinely screened for thyroid problems on blood tests, but are not often routinely tested for coeliac disease.

It is recommended (by groups like NICE who advise on UK health issues) that coeliac disease should be looked for in individuals who are at increased risk. If there are no symptoms that relate to the bowel it is sometimes difficult to know why we should be doing the test, and we might need to discuss the pros and cons of doing the test with you - we are happy to do this and contact details are below. There are two approaches and we wanted to explain how we would go about doing testing.

Coeliac disease

Coeliac disease is a common autoimmune condition in individuals who have a genetic predisposition to it. It is an immune reaction to gluten, a protein found in wheat, rye and barley. People might have no symptoms at all but still may have the condition. Typical symptoms can include bloating, gassiness, abdominal pain, loose stools and constipation and can develop at any age. It damages the lining of the small intestine and causes poor absorption of nutrients and can cause anaemia and thinning of the bones. These problems and other longer term risks in children are completely reversible on a gluten free diet (which is the treatment for the condition).

What we are proposing

We would like to invite your child to have a blood test to screen for this condition. The blood test is to determine whether your child carries the risk factor gene for coeliac

disease and what their standard coeliac blood test shows - these are tested at the same time.

People with coeliac disease have a predisposing genetic factor called DQ2 or DQ8 which almost all patients (over 99%) have. It is a common finding in the general population (approx. 30%) but only 1% of the population have coeliac disease. Being positive for this genetic factor therefore means there is a risk of coeliac disease, but most will not have it. If your child carries this factor, we would continue to offer a coeliac antibody test every few years.

If your child does not carry the genetic factors DQ2 or DQ8, it is extremely unlikely that they would ever develop coeliac disease. This means that they would not require to be screened for coeliac disease again in the future.

By doing this test, we are hoping to improve the service we are providing to children with Down syndrome and their families and to help pick up the condition earlier and avoid problems in the future.

Further information is available on the **DSMIG website** (www.dsmig.org.uk) and in the patient information sections on the **Coeliac UK website** (www.coeliac.org.uk), **BSPGHAN coeliac guideline** and in the **NICE NG20 quick reference guide** (www.nice.org.uk/guidance/ng20).

If you have any questions and wish to discuss this further with us, our contact details are below. If you wish to arrange testing, please contact **Dr Timothy Lewis**, or your own community child health paediatrician, who will be happy to help.

We will follow up this letter with a telephone call as a courtesy.

Yours sincerely

Electronically checked and signed by

Dr Timothy Lewis Tel: 0131 xxxxxxxxx
Paediatric Registrar

Dr Sarah Clegg Tel: 0131 xxxxxxxxx
Consultant Paediatrician

Dr Peter Gillett Tel: 0131 xxxxxxxxx
Paediatric Gastroenterologist

NOTES FOR THE SENDOUT LETTER AND OUR PHONE DISCUSSIONS ……..

Reading it now, we left a lot to discuss at the phone call. You may wish to be more descriptive in your letter.

This was clearly an 'opt in'. There was absolutely no compulsion to agree but we did follow up on the letter.

The phone call chat – ''we sent you a letter a few weeks ago, and wondered what you thought?''. Click……..OR……

Options / things discussed during the chat were…..

- Do nothing – never tested and don't want tested. W and W (watch and wait). ''Come back to us or your Comm Paeds team if there are any issues / you change your mind''. No letter sent (there maybe should've been).
- We had Standard serology done before and normal and we are OK thanks. Plan would be to Repeat at some point or just watch and wait.
- DQ only (maybe tested before or never tested) please as we would like to do that first if the chances are good that it is negative or standard serology has been negative in the past and we know we are not 'currently coeliac.
- Both DQ and serology / maybe repeat serology if, on discussion they wanted both, it had been more than three years (ad hoc timeframe) or there were symptoms possibly suggestive of CD. My own bias was to advise this if they had another AI condition (we have i think two children with T1DM, clearly a significant number with thyroid issues).

DQ Results…….

- Negative . letter detailing and offer to discuss. STOP screening
- Positive, they got a copy of the formal report and a written explanation by me of what it meant. If they had serology done ever / at the same time, it was helpful to be encouraging that the risk is low, lifetime, of course. The letter never really raises worries re 'high' risk, as it's 'all relative', and relatively few would ever become coeliac anyway, even from a 'pool' of positive DQ types, but clearly that's another reason for 'risk stratifying'.
- To put in context, In the 'regular population, say we look at 100% being tested ie ALSPAC , then 1% are coeliac – BUT if you sift out the negatives (ie 60%) you are left with 40% of the regular population who are at risk – of those 40%, 3 % will be coeliac 'ever'. This compares to 1% of the 100%. Hope that makes sense. I will try to work out a similar thing for DS ie if 60% carry the permissibility gene…….