DiGeorge Syndrome (Review and National Health Service Duty)

05 June 2018

Volume 642

Motion for leave to bring in a Bill (Standing Order No. 23)

2.29m

David Duguid (Banff and Buchan) (Con)

I beg to move,

That leave be given to bring in a Bill to require the Secretary of State to conduct a review into DiGeorge (22q11 deletion) syndrome; to require the National Health Service to publish a strategy after the review is complete; and for connected purposes.

This Bill would, as its long title says, place a duty on the Secretary of State and the Department of Health and Social Care, in consultation with the national health service and key stakeholders, to conduct a review into the diagnosis and treatment of DiGeorge syndrome. Those stakeholders should include sufferers from DiGeorge syndrome and their families. The Bill would also require the NHS to develop and publish a strategy for DiGeorge syndrome after that review had concluded.

For the purposes of the Bill, I have referred to this condition as "DiGeorge syndrome" with "22q11.2 deletion" in parentheses, as "DiGeorge syndrome" is the name most commonly used by the NHS. However, because the signs and symptoms of this syndrome are so varied, different groupings of its features have historically been regarded as separate conditions under a variety of different names. Those conditions are now understood to be presentations of a single syndrome with the universally accepted nomenclature of "22q11.2 deletion syndrome". For brevity, Members will be glad to hear, I will refer to the condition as "22q" from now on.

22q is a genetic disorder caused by the deletion of between 30 and 40 genes in the middle of chromosome 22 at a location known as 22q11.2—hence the name. 22q is often referred to as a rare genetic condition. However, as the second most common chromosomal syndrome after the much more familiar Down's syndrome, it is unfortunately not as rare as the little-known name might suggest. Estimates have ranged between one in 4,000 and one in 2,000 live births, although due to the lack of familiarity with the condition, that is expected to be an underestimate. In fact, recent estimates suggest that it is as common as one in 1,000. 22q is often described as "the most common genetic syndrome you have never heard of."

Everyone with 22q is affected differently. Most children with the condition survive to adulthood and enjoy a relatively normal lifespan and an independent life. However, 22q can lead to a range of health issues that can affect quality of life and even shorten lifespan. Congenital heart disease is a common

concern, as well as defects in the palate and a range of learning difficulties. Worryingly, children who are born with 22q and present with learning difficulties are often misdiagnosed as being on the autistic spectrum. Studies have shown that although some of the developmental symptoms may be similar, the causes of the symptoms are quite different. That common misdiagnosis often leads to 22q patients receiving the wrong kind of care and support, with potentially disastrous effects.

Because the different treatments offered can be as varied as the root causes themselves, children with 22q often develop other medical issues, particularly concerning mental health, that could otherwise have been avoided if diagnosed correctly. Mental health issues are very common among patients living with 22q, particularly if diagnosis is missed early on. In 22q patients, mental health issues often present themselves at a much earlier age than in the general population. Schizophrenia, for example, normally has an occurrence of about 1%, whereas in the 22q population it is estimated at closer to 25%.

When I first became aware of the condition, one of the concerns that surprised me the most was the relative lack of familiarity of 22q among not only parents and educators but those in the medical profession, particularly in general practice. Because of that, many symptoms go unnoticed until they have already progressed considerably. This blind spot not only means that the NHS incurs additional costs in the long term but has a terrible impact on the patient, their family and their carers, as well as affecting employment, quality of life and mental wellbeing. Early detection of 22q provides the opportunity for early intervention and management of the condition, which can significantly improve the quality of life of patients and their families.

At this point I would like to pay tribute to the charity Max Appeal, which does a huge amount of very valuable work on 22q. It has produced a paper that argues for the introduction of a screening programme for 22q that provides earlier diagnosis of affected individuals, in turn reducing morbidity and possibly even mortality. For a relatively small cost, such a screening programme would help to keep overall healthcare costs down due to the early detection of potential related issues and the complications that I have mentioned.

22q can be detected through a simple heel-prick test that the NHS already offers to babies around five days after birth. The test entails pricking the baby's heel and collecting four drops of blood that are then tested for a total of nine conditions, including the more well-known sickle cell disease and cystic fibrosis. At the moment, no screening for 22q is performed routinely on the blood collected through heel-prick testing. Any blood tests to check for 22q are done only to confirm a diagnosis when another symptom prompts clinicians to do so, such as in the case of a cleft palate or heart defect. Clearly, that approach cannot be relied on to catch all cases, partly due to the surprisingly low awareness of the condition among GPs and other medical professionals not specialising in the field of rare genetic conditions.

The UK national screening committee does not currently have a recommendation on screening for 22q, although some work has been done on it in the United States. For example, a 2014 study published in

the journal *Clinical Chemistry* found that blood spot tests would be a promising approach for newborn screening for 22q. A 2017 study in the *Journal of Clinical Immunology* looked primarily at screening for severe combined immunodeficiency, or SCID. That study discussed the potential introduction of newborn blood spot testing for 22q. The authors of the study concluded:

"Assays which screen for 22q11.2 Deletion Syndrome using dried blood spots have been developed and proven to be effective and efficient...Population-based studies should be completed to demonstrate the efficacy of these assays on a larger scale...However, the clinical characteristics, diagnosis, management, and treatment of 22q11.2 Deletion Syndrome have been shown to meet the criteria for new-born screening programs and support the need for earlier diagnosis."

Screening for SCID is currently being trialled in the UK, and it is believed that it could be extended to a "two birds with one stone" approach by trialling screening for 22q. As I mentioned, Max Appeal has produced a paper that argues the case for that. I hope that the Secretary of State and the relevant Ministers at the Department will consider reading it.

I know that Ministers and fellow Members will be as concerned as I was to learn of the struggle facing 22q patients and their families. It is for that reason that I am asking the House that leave be given to bring in a Bill to require the Secretary of State and the Department for Health and Social Care to conduct a review into 22q, and to require the NHS to publish a strategy after that review is complete. Whatever the outcome, that would be of real value and of real reassurance to the thousands of families around the UK who are affected. There is so much that we can do to help those families in a sensible, proportionate and cost-effective fashion.

Finally, I would like to invite the Secretary of State, his Ministers at the Department and any hon. Members who would like to hear more about this issue to attend the next meeting of the all-party parliamentary group on 22q11 syndrome, of which I am the chair, on 26 June. I know that Max Appeal and the families that it represents, who are spread across the country in many colleagues' constituencies, would be very grateful to see them there.

Question put and agreed to.

Ordered,

That David Duguid, Heidi Allen, Mr Robert Goodwill, Alex Sobel, Norman Lamb, Jack Lopresti, Vicky Ford, Melanie Onn and Jim Shannon present the Bill.

David Duguid accordingly presented the Bill.

Bill read the First time; to be read a Second time on Friday 26 October and to be printed (Bill 218).