



**News release: 5 June 2018**

## **David Duguid MP calls for routine screening of rare genetic disease**

**Today (5 June 2018):** Scottish Conservative MP and Chair of the All Party Parliamentary Group (APPG) on 22q 11 David Duguid will propose a Bill to Parliament, seeking the introduction of routine screening for 22q 11 so it can be diagnosed and treated earlier.

### **Julie Wootton, Founder of Max Appeal, in memory of her son Max said:**

“22q 11, is the second most common chromosomal syndrome after Downs Syndrome that affects thousands of children every year but is frequently misdiagnosed. This is a vital first step to include 22q screening into the tests that currently exist. We know that delays in diagnosis not only have adverse affects on the child and family but also treatment costs for the NHS are likely to be less efficient in the long term.”

The effects of 22q 11 are wide ranging, including congenital heart disease, defects of the palate and a range of learning difficulties which lead to the disease being misdiagnosed. This results in patients receiving the wrong kind of care and support which may impact on their quality of life, mental health and employment opportunities.

Mr Duguid was made aware of 22q 11 through his constituent, Louise Henderson, who raised the issue with him at a surgery. He is aware of other members of his constituency who have the condition. He has since worked closely with the charity Max Appeal and the APPG to call for earlier and routine screening by using heel prick tests that are given to 5-day old babies and screening of Severe Combined Immunodeficiency (SCID) which is currently being trialed in the UK.

### **Commenting, the MP for Banff and Buchan David Duguid said:**

“I am calling for the introduction of a screening programme so there is a better chance of catching this rare genetic disease. At present it’s only detected when another symptom prompts doctors to examine people in more detail. This approach cannot be relied upon. 22q screening can be incorporated into tests that already exist, such as heel prick tests that are given to new born babies. This ensures the right diagnosis and the right kind of care can be given. By proposing this Bill, I hope to raise awareness of this condition, and ensure a review takes place in the hope of bringing in routine screening.”

**Ends**

### **Notes to editors:**

The story of Mr Duguid’s constituent, Louise Henderson, is attached. Link to the Max Appeal website is [here](#)