**Minutes: APPG 22q11 Syndrome Meeting – 13th September 2023**

**Attendees**

* David Duguid MP (DD)
* Julie Wootton (Max Appeal) (JW)
* Paul Wootton (Max Appeal) (PW)
* Samantha Callan (Lord Farmer’s Office) (SC)
* Elizabeth Barker (EB)
* Jonathan Tucker (David Duguid’s Office) (JT)

**Apologies**

* Baroness Eaton DBE DL (ME)

1. Attendees introduced themselves.
2. EB begins by talking of her experiences with the heel prick test which currently does not pick up 22q syndrome, a diagnosis of which would have made sense of the many conditions which began to emerge early in her son's life. He had Hirschsprung’s disease which was discovered at 10 days old. He also had narrow airways and resultingly could not cry properly until they were operated on. He also struggled to put on weight and had low muscle tone. It was later found that his vocal cords were fused together and he had a minor heart defect. Without a proper explanation, EB said that she and her husband began to blame themselves as parents.
3. EB said that it took her son over 1 year to be diagnosed with 22q – still earlier than the average diagnosis.
4. EB felt relieved when she found out her son’s diagnosis. It gave her relief that she was not to blame. It also gave her hope that a solution could be sought.
5. DD suggested that in future meetings an affected parent/person could briefly share their own experience at the beginning.
6. EB pointed out a reason for misdiagnosis, or lack thereof, is parents going to different professionals who diagnose based on symptoms that are only within the purview of their expertise. There is a necessity to diagnose from a holistic perspective.
7. DD asks JW whether there are any signs that awareness of 22q in the medical community is improving. JW said there are - she knows a University Lecturer who can attest that more of his students, when asked, know about 22q.
8. PW said that it is often a matter of finding the right consultant – someone who knows about 22q and is aware of how it manifests.
9. JW mentioned that different 22q traits can emerge and develop at different times in a child’s life. They do not necessarily manifest all at the same time. This makes it harder to diagnose 22q.
10. JW affirms that new born screening is the definitive way of ensuring whether someone has 22q or not. 22q can often blend in with other conditions, such as Downs Syndrome and the only way of distinguishing 22q is through screening.
11. DD suggests making representations by writing a letter as an APPG.
12. JW said that there is no coherent treatment pathway and one needs to be put in place. DD made the point that, given that there is no specialty treatment for 22q, one cannot be established. Would the NHS be interested in identifying 22q if they do not have a bespoke treatment in place?
13. SC queried whether short-term intervention may help particularly focusing on the mental health of parents going through the process of raising a child with 22q, in respect of dealing with the effects and battling with various agencies about necessary provisions. It is often a stressful and traumatic experience with risk factors involved, including suicide. JW adds that it is potentially a marriage-breaker.
14. PW suggested maybe finding a 22q champion.
15. SC suggested DD puts in the Private Members Bill ballot. DD always does but, so far, to no avail.
16. JW mentioned that she was recently contacted by the Police about whether any offenders may have 22q. In Northern Ireland prisons are going through their inmates to test whether they have 22q.
17. DD spoke about the National Screening Committee (NSC) will need to be shown evidence that screen testing will make improvements. JW suggested that testing people can be done in an order, particularly those whose situation are easiest to treat more quickly should be tested first. This may then provide the basis of proof needed. JW then pointed out that the number of people with 22q are great – the NSC/NHS may not want to commit to.
18. JW also touched on adult care and school provisions. She said a big issue is 22q children having to prove their learning disability. They are classed as having learning difficulties – meaning their challenges can be resolved through established means.

**Actions**

* Status Completed - DD makes representation as APPG to National Screening Committee before 25th September deadline.
* Status Completed - JW drafts wording of DD’s representation beforehand.