**Minutes: APPG 22q11 Syndrome Meeting – 18th July 2023**

**Attendees**

* David Duguid MP – Chairman (DD)
* Liz Twist MP (LT) – Chair of Rare & Genetic Disorders APPG
* Julie Wootton (Max Appeal) (JW)
* Paul Wootton (Max Appeal) (PW)
* Gill Croft (Mother who has son with 22q DS) (GC)
* Samantha Callan (Lord Farmer’s staff) (SC)
* Jonathan Tucker (JT)

**Apologies**

* Baroness Eaton DBE DL (ME)
* Carla Atwood (Max Appeal)
* Mandy Sanderson (Max Appeal)

1. Attendees introduced themselves.

2. Committee went through previous actions set at last AGM:

* DD submitted two Written Parliamentary Questions (WPQ) regarding blood screen tests for new-borns and inserting 22q11 guidance into the Down Syndrome Act.
* DD then submitted two letters to DHSC Ministers based on unsatisfactory answers to his WPQ’s. APPG went through the answers that DD and MF received from DHSC which were disappointing.
* DD has not applied for debate either for Westminster Hall debate or Adjournment debate.

2. LT contributed with her knowledge from the Rare and Genetic Disorders APPG about the matter of new-born screening (heel prick test). She also raised that Helen Whately is now the minister responsible for rare genetic diseases.

3. The New-Born Screening Committee was discussed and it was agreed that the way it addresses applications and additions to the list appears ad hoc and gives insufficient regard to international findings particularly around frequency within the population. Recent proportional studies from USA suggest that upwards of 30,000 people could have 22q but there are only 2,500 members of Max Appeal, which still makes it the largest charity globally for the condition, particularly with per capita considerations.

4. Max Appeal has been in discussion with pharma company, Natera, and has applied for a grant in the region of £12,000 due to their programme of increasing awareness of the Panorama test that can identify foetal DNA from maternal blood samples and so has no risk to the baby. JW highlighted that this is an emerging technology and that presently the figures for false positive results is disturbing and that a positive result then has to be followed up with either CVS or amniocentesis for a conclusive result and both of those carry risks to the unborn baby, but this technology may well improve and become more widespread and reliable in the future and is, therefore, important to recognise. DD felt that caution should be shown and this was agreed by all.

5. DD highlighted that Business Questions could be useful for raising awareness for 22q, if not for trying to secure a debate.

6. JW summarised the issues faced by parents with the special education needs provision; that being the gaps in flowing through transition stages to senior schools, colleges and higher education.

7. JW stated that Republic of Ireland is ten years ahead of UK for advancing 22q matters. On Saturday 18th/Sunday 19th November there is a European conference on 22q in Dublin: <https://22q11europe.org/22q11-european-conference-in-dublin/> .

8. JW and SC begin talking about gaps in provision for education for SEN children going into further education. JW highlighted that education provision has to be meticulously planned for the entire duration of education, otherwise one hiccup could lead to SEN children falling considerably behind. There was also talk about whether some schools are willing to take on this responsibility.

9. GC begins talking about her own story with her son. Her son is 13 years old and was diagnosed 6 years ago. His school could not accommodate him fully despite insisting that they could. She said that his learning disabilities were the cause of his behaviour.

10. JW recounted recent conversation with a mum of an 18 year old daughter, Joanne Beard, and the hurdles she has encountered with regard to meeting attendance criteria and that her daughter’s ill health precludes her from achieving the requirement and that, despite achieving academic standards, her place from September had been withdrawn. This clearly is unfair and precludes her from achieving her potential and aspirations as an adult.

11. GC highlighted the problem of awareness of 22q amongst health care professionals, saying that her son’s diagnosis was not understood or really recognised at all. Her son is adopted, along with his brother, and this extra resources both financial and input from a medical team led to his diagnosis a few years ago. Her and her husband have now managed to secure a SEN placement for her son but it has been a struggle over many years and there is still on outstanding issue about funding to overcome which has meant he has not been able to attend the new setting for the usual transition process which is to his detriment.

It was agreed that, sadly, these issues are not unusual and have been aired on many occasions by many families. There is still a lot to be done.

12. JW said that as 22q is a spectrum of symptoms, it is worth exploring avenues to differentiate it from other conditions, such as autism. It would be worth getting tested for autism to distinguish.

13. DD and JW spoke about 22q’s prevalence, highlighting that Cystic Fibrosis is a third as prevalent. JW mentioned that 1800 people have been diagnosed with 22q.

14. JW spoke about how there is an ongoing pilot study into the genetic testing of 22q. There is a Michigan study on this to be looked up.

15. Everybody discussed the possibility of doing a documentary in mainstream channels, for example Channel 4 or BBC Panorama. JW suggested the documentary could focus on the issue of the volume of false positives. JW also mentioned there is an insightful TV show with David Tennant on raising a child with learning disabilities as DYRK1A Syndrome – There She Goes. DD then mentioned Retained Primitive Reflexes which JT will share with JW.

16. JW said we were missing a trick in not speaking to enough people about 22q. Channels and forums would need to be explored and their media could be advertised.

17. DD suggested that instead focus on getting more people engaged and aware of 22q rather than seeking a final outcome at this stage. JT suggested hosting a drop in after Conference recess that DD’s office would advertise, but also Max Appeal members email their respective MP’s to urge them to attend. DD said it would be the sort of event where members would come, leaflets with food and speeches. PW then suggested that this could coincide with the relaunch of the consensus document. JW said that it may be possible to have this event sponsored by Natera – a pharmaceutical company.

18.  No further matters were raised, and David Duguid MP declared the meeting closed.

**Actions**

* DD to look into raising further written questions about the Down Syndrome Act RE: why people with that specific condition and the learning difficulties associated with it should be dealt with differently to a child/young person with another genetic condition that causes them to have an intellectual disability.
* JT and DD plan date and time for next 22q meeting for W/C 11th September.
* DD consider with JT raising 22q through parliamentary channels, either through Business Questions or a backbench debate – Westminster Hall or Adjournment debate about heel-prick test.
* DD looks into TV show, There She Goes.
* JT shares information with JW on Retained Primitive Reflexes.
* DD, JT and JW agree on a date and function for a 22q event in November.
* Max Appeal to further investigate the 100,000 Genomics project and continue to press for 22q to be added to the pilot.
* DD to continue to seek sponsorship from an SNP to hold a reception in Holyrood.
* Max Appeal to encourage its membership to write to their MP for when an event is decided.
* Max Appeal to try to identify how many people affected in each constituency from its database and estimated of prevalence to highlight that the vast majority of affected individuals remain undiagnosed.
* JW to provide details of Michigan study when available