22q11DS All-Party Parliamentary Group Meeting
Tuesday 2nd December 2014
Committee Room 20, House of Commons
10.30am ~ 11.30am

Draft Minutes

Present:
Jack Lopresti MP
Margot James MP
Paul Uppal MP
Marcus Jones MP
Alastair Burt MP
Selina Short – JL Office Manager
Julie Wootton - Max Appeal
Paul Wootton – Max Appeal
Claire Hennessey – Max Appeal
Carla Attwood – Max Appeal
Gillian Cassidy – 22qNI
Keith Markham – 22Crew
Dr Alex Habel – Medical Expert
Dr Clodagh Murphy – Medical Expert

Apologies:
Nigel Adams MP
Heather Wheeler MP
Debbie Abrahams MP
Graham Evans MP
Kevin Hopkins MP
Peter Wishart MP
Anne McGuire MP
Jack Berry MP
Robert Walker MP
Andrew Bridgend MP
Roger Perkins – Govia Thameslink

Item
1. Welcome and Introduction by Jack Lopresti MP Jack Lopresti opened the meeting and stated his pleasure that the required number of sponsoring MPs had been achieved so that this important All-party Parliamentary Group could be established.
2. Apologies Selina Short noted apologies as above.
3. Election of officers Jack Lopresti - chair proposed by Paul Uppal seconded by Marcus Jones

Margot James – Vice Chair
proposed by Jack Lopresti
seconded by Paul Uppal

Paul Uppal – vice chair
proposed by Nigel Adams
seconded by Margot James

Paul Wootton agreed to take minutes of this meeting and Julie Wootton confirmed that Max Appeal would act as secretariat to the APPG.

4. Introduction to 22qDS by Dr Alex Habel

Dr Alex Habel, retired consultant paediatrician from Great Ormond Street Hospital and lead consultant for specialist 22q clinic, spoke about the condition.

He explained that 22q11DS is a genetic condition that affects people in many varied ways. 85% of people affected are because of a random mutation that causes genes to be lost or copied on the 22nd chromosome. 70% or more of people with the condition suffer from congenital heart defects, palate problems, learning difficulties and psychiatric and mental health problems. The current estimated prevalence is 35,000 people in the UK with the numbers increasing as people survive heart surgery. The average IQ is 70, particular difficulties are with maths and numbers, whereas language and learning are less of an issue.

The age of presentation is important as is early screening and genetic counselling for parents. Co-ordinated support is vital for the individual and the family to ensure access to and provision of joined up services. Other issues experienced include socialisation, and vulnerability to exploitation and bullying.

Issues encountered by adults with the condition were discussed, including low self-confidence and poor employment possibilities.

Alistair Burt questioned whether there were any geographical or ethnic bias, Alex Habel responded that there weren't.

Marcus Jones declared that he may have a personal interest in the condition and requested to speak to Alex Habel privately after the meeting. He questioned the severity of the condition and how many of the issues would be presented. Alex responded that the severity of the condition varies considerably and that there could be many combination of issues and presenting features.

Margot James enquired as to how easy it was to test for 22q. Alex Habel described the types of genetic tests and DNA probes and how this has developed over the years so that now 50,000 DNA probes are used in a full array test. Alex Habel then mentioned that Julie Wootton had worked with a team in the USA to establish neo-natal testing in the USA as a standard test. Julie Wootton explained that pulse-oximetry screening was the norm in the USA but was still being debated by the NHS as to whether it should be adopted in the UK. Julie Wootton explained that a blood spot is already taken at birth from every baby and so it is a matter of what is tested using that blood. Julie Wootton and Alex Habel agreed that this encountered many ethical problems.

A briefing document had previously been prepared by Selina in conjunction with Max Appeal to provide members with an overview and this had been circulated prior to the meeting by Selina. (Appendix 1)

5. Living with a child with 22q by Carla Attwood

Carla's daughter, Georgina, presented with speech concerns at 22 months old, and was referred to a speech and language therapist who advised on the possibility of having 22q deletion. Grommets were fitted to try to improve her speech. Georgina was eventually diagnosed with 22qDS at 3 years of age and had an array of tests completed over the subsequent 8 months. Carla emphasised the benefit the benefit of having the specialist multi-disciplinary clinic at Frenchay Hospital (that has now moved to Bristol Royal Infirmary) and key milestone support. Georgina required two palate operations but her speech is now very clear and the hypernasality had been cured.

Margot James raised questions in relation to learning difficulties. Carla responded that her daughter received weekly speech and language support and 10 minutes a day teaching support and that Georgina attends a private school because Carla had been advised that she would receive no support within a state school environment.

Keith Markham expressed his experiences with his daughter and mirrored Carla's comments that he too had been fortunate to receive good care and support both medically and educationally within his borough.

Gillian Cassidy spoke about hr experiences as a parent with a child with 22q in Northern Ireland. She described the difficulties parents who have 22q11DS themselves encounter.

Alistair Burt raised a question on the impact on medical professionals. Dr Habel responded that part of the issue was continued lack of resources and that he had sought to improve awareness of the condition through the National Consensus Document of the Diagnosis and Management of 22q11DS
http://www.maxappeal.org.uk/knowledge/consensus_document/content.asp?s=44&p=67
and through an article published in the European Journal of Pediatrics titled "Towards a safety net for management of 22q112 deletion syndrome: guidelines for our times."
http://link.springer.com/article/10.1007%2Fs00431-013-2240-z But also acknowledged that awareness needed to be improved at undergraduate level for medical and education professionals alike.

Claire Hennessey commented that it was important for educational and other professionals to be aware of 22q11DS as they had day to day interaction with children and young people with the condition and so they were instrumental to their development.

6. APPG Aims for This Parliament

It was agreed that the main aim of the APPG was to raise awareness of the condition by way of an event at the end of February (date to be agreed) between 4pm and 6pm, it is intended that the Health Minister be present at the event and to respond at the meeting.

Action: Selina to book a room, possibly members' dining room a or b.
Action: Margot James would request an adjournment debate in the House.

Action: Max Appeal to encourage 22q families to write to their MP informing them of the APPG and to highlight the need for awareness of the condition.
Max Appeal to issue press release. (Appendix 2).

7. DNM

Tuesday 13th January 2015

Action: Selina to confirm.

8. AOB

None

Jack Lopresti closed the meeting and called for photos to be taken.

Appendix 1

All-Party Parliamentary Group for 22q11 Syndrome

Briefing Document for members of the APPG from Jack Lopresti MP

Why?

- A constituent's daughter suffers from this syndrome and it has resulted in her having Learning Difficulties. The parents would like the syndrome recognised and tested for by the NHS because it will have on going effects throughout her life.
- 22q11 syndrome is estimated to effect up to 35,000 people in the UK.
- Detection rate is very low, so most people do not get the help they need.
- Max Appeal is a charity for individuals and families affected by 22q11 syndrome.

What is the Syndrome?

There are about 40 missing genes on part of the long (q) arm of the 22nd Chromosome.

Deficiency may result in:

- Palate defects
- Cardiac abnormalities from birth
- Many endocrine (gland and hormone) issues
- Immunity problems and auto-immune disorders
- Learning difficulties
- Attention deficit
- Very high prevalence of severe mental health disorders

A National Consensus Document written by a many of eminent clinicians with over 40 contributors in total, has been published by the Max Appeal. The group was chaired by Dr Rick Herriot, consultant immunologist Aberdeen Royal Infirmary, and the document was edited by Dr Jeremy Allgrove, consultant paediatric endocrinologist at Barts and The London NHS Trust.

Aims

- Max appeal is looking for the NHS to be able to compile **a comprehensive and universally agreed lifelong care plan for people with 22q11 syndrome.**
- The number of people suffering with the syndrome is growing because of better medical care and as this is most often not an inherited genetic syndrome it is currently not possible to predict who is affected or will have a child with the condition.
- Therefore increasingly important that once diagnosed a multi-disciplinary team approach will be required for most people with the syndrome.
- Treatment and support for people with Syndrome can be improved if a joined up approach is followed by Healthcare professionals.
- APPG will bring the 22q11 syndrome to the attention of policy makers.

Appendix 2

News release: 3rd December 2014

MP's join forces to support children with 22q11 Syndrome

All Party Parliamentary Group
set up today for Bristol and Stourbridge based children's charity

Today (2 December 2014) Jack Lopresti MP chaired the first meeting to set up an All Party Parliamentary Group [APPG] for Bristol-based Max Appeal. The charity supports those affected by 22q11 Syndrome - a complex condition that is often undiagnosed and leads to lifelong struggles for those affected.

Jack has set up the APPG to raise awareness of this medical condition after meeting one of his constituents who has a daughter with the condition. Nigel Adams MP is the Vice Chair and Margot James MP – a long term supporter of Max Appeal – is Treasurer.

These conditions affect health and quality of life from birth through infancy and childhood into adult life with over 180 physical, functional and psychological associations reported. The complexity of 22q11 frequently leads to clinical confusion and a delay in diagnosis often by years.

At the first meeting parents gave their personal experience living with a children and young adults with 22q11 Syndrome. Dr Alex Habel, retired Consultant Paediatrician Great Ormond Street Hospital, discussed how new genetic technology is helping identify those with 22q11 syndrome, both deletion and duplication.

He says "It is the key to improving these individual's lives by early and anticipatory delivery of care. From conception onwards it affects many body systems, including the heart, palate, immune system, behaviours and mental health. Care is therefore best provided by a multidisciplinary team throughout the life cycle. Education about early recognition of 22q11 syndrome should extend to all professional disciplines who specialise in delivery of health and psychological services, and education, to children".

Jack Lopresti MP says; 'I set up this APPG to raise awareness about this syndrome that is estimated to affect up to 35,000 people in the UK. Currently the detection rate is very low which means many people are not getting the help they need. I am delighted that so many of my colleagues have joined this APPG.'

Julie Wootton, chair of trustees, Max Appeal, says:
"We are delighted to be launching this APPG and are incredibly grateful to Jack Lopresti MP and the MP's putting their weight behind it. We hope that this will help to raise both awareness and understanding of the needs of these families, their children and adults living with the condition."

- Ends -

Further information:
Emma Pelling, emma@pellingpr.co.uk, t. 020 7624 7533 / 07958 558172
www.maxappeal.co.uk

Notes to Editors

1. Case studies are available for interview, please call the numbers above to make arrangements.

2. Max Appeal is a small national charity, run almost exclusively by a group of dedicated volunteers backed up with a knowledgeable panel of medical professionals**.** The charity was inspired by Max who sadly had a short life. Today, Max Appeal is a charity with a vibrant life of its own. The charity's mission is to ensure that every person with 22q11.2 deletion lives an independent and prosperous a life as possible