

## Virtual Cross Party Group on Rare, Genetic & Undiagnosed Conditions

21 September 2020, 2.30 - 4.00pm

Theme: Impact of COVID 10 on Rare, Genetic & Undiagnosed Community in Wales

Present:

Angela Burn MS - Chair  
Emma Hughes (Genetic Alliance UK) - Secretariat  
Mike Bryan (Angela Burns' office)  
Nick Meade (Genetic Alliance UK)  
Sophie Peet (Genetic Alliance UK)  
Rachael Humphreys (Patient affected by Behcets)  
Sondra Butterworth (Patient/ Rare Disease Researcher)  
Sylvia Evans (World Orphan Rare Lung Disease - WORLD)  
Alan Thomas (Ataxia & Me)  
Lesley Harrison (AKU Society)  
Tony Thornburn (Behcets UK)  
Menai Owen-Jones (Pituitary Foundation)  
Marie James (Tuberous Sclerosis Association/ Parent)  
Alison May (Cardiff Friends of Sickle Cell & Thalassaemia Supporter)  
Grace Paget (Cystic Fibrosis Trust)  
Michael Cain (HSP Support Group)  
Samuel Chawner (Cardiff University)  
Tracy Blockwell (Patient - Ehlers Danlos Syndrome)  
Tony Crowhurst (Disability Advice Project)  
Ryland Doyle (Mike Hedges' office)  
Lewis Troke (CLL Support)  
Graham Shortland (Rare Disease Implementation Group)  
Tessa Hughes (Map BioPharma)  
Joanne Ferris (ABPI Wales)

**1. Annual General Meeting** - Nominate and vote for Chair (Angela Burns MS) and Secretariat (Emma Hughes, Genetic Alliance UK). Both will stand again.  
No objections to Chair or Secretariat, both roles to continue.

### **2. Introduction from Chair, Angela Burns MS**

Angela Burns MS: Thanks for allowing continuation of role. Angela won't stand after Senedd elections in May. Angela will advocate and push for replacement from MS colleagues and interested parties to continue valuable work of group.

### **3. Rachael Humphreys – presenting on her personal experience of 'Surviving shielding'**

Key points from presentation

## **Behcets Disease**

- Rachael has a rare auto-inflammatory vasculitis disease - Behcets and has a more aggressive form of the condition.
- Treatment is targeted at the symptoms of the condition. The more aggressive the symptoms, the more aggressive the treatment regime. Many patients take immunosuppressants.

## **COVID 19 & lockdown/ shielding experience**

- 2020 felt like surviving an invisible threat.
- Listened to the news with growing anxiety/ new language in society, 'lockdown', 'shielding'.
- Real sense of not being looked after or cared for before lockdown as deaths being reported in an insensitive way – People dying are “only” those with underlying health conditions – a real sense of it therefore didn't matter/ feeling of vulnerability in rare disease community
- Lockdown felt like we were being protected by people who didn't know us.
- Delay of announcements or saying there “would be an announcement tomorrow” – increases anxiety
- Had to limit access to news – too overwhelming
- Categories for shielding were not clear - were people with Behcets clinically vulnerable or not? Do they need to shield or just practice strict social distancing?
- Keeping physically and mentally well was a big challenge. Advised by GP to shield elsewhere when daughter became unwell as wouldn't survive if Rachael caught it. Reality of COVID 19 when you have a rare disease and are immunosuppressed. Felt completely out of control, panic attacks and high anxiety. Spent 2 weeks away from family shielding. Focused on mental health and wellbeing through mindfulness and wellbeing activities.

## **Impact of COVID 19 on care/ treatment**

- Shielding letter - don't leave the house. Have food and medicines delivered.
- Affect with care - rheumatology appointment changed to online. Ophthalmology appointment has been rescheduled twice and doubts this will happen.
- Many patients had medication switched from Infliximab to a self-injection so this could be administered at home. Patients struggled to get hold of specialists.
- Difficult to follow restrictions set out in letter with 2 young children. Can't follow the shielding advice to the letter – looked at risk and how to reduce it.

- Prioritised mental health -decided to get out of house and go for runs. A lot of people advised by GP and specialist that they should leave their house to take some form of exercise. This had extremely positive impact - anxiety reduced so could reduce anxiety medication.
- Definitely a survivor rather than a thriving during this time. Home became safe haven and worked from home in adapted role.
- Not everyone is cautious or respectful of keeping distance because they don't have same underlying health conditions.
- Updated risk assessment/ navigating back into work environment was difficult.
- Worry and anxiety growing again - in local lockdown in Caerphilly but very few changes to how things are functioning i.e. school/ work. Added threat son may bring coronavirus home.
- Confusion over decisions
- If asked to shield again - being able to leave house for exercise should be allowed and those living alone should be able to form a bubble to reduce loneliness

#### **Concerns raised by other patients/ families**

- Main questions Rachael received from groups and other patients/ families:

-Will we have to shield again?

-Is it safe for us to go out and exercise?

#### **What next?**

-Need vaccine to feel safe/ protected – will the vaccine be live or inactive?

-Will those who are extremely clinically vulnerable be prioritised?

-How effective will the vaccine be?

-Will people need to shield again?

#### **4. Discussion following Rachael's presentation**

- **Tracy Blockwell** – some support measures put in place are not suitable for those with disabilities, particularly those who live on their own. Example: Online shopping doesn't work because she can't lift the shopping. Can't get help as shielding. Very fearful of another lockdown as has to put herself at risk. Concerned as going into another lockdown. For a lot of people with disabilities - managing the best way they can.
- **Graham Shortland:** Written to Chief Medical Officer about impact on rare disease community and key points - will be noting down points as people are speaking.

**ACTION: Angela Burns MS to write to Vaughan Gething following CPG highlighting experiences of COVID 19**

- **Angela Burns MS:** Cannot eradicate all risk - need to try and minimise it. Mental health issues of having to shield and being away from your family is devastating.
- **Graham Shortland:** Rachael raised a good point. One area of influencing policy is around vaccinations - prioritise access for those who are at greater risk,
- **Tony Crowhurst** - Point made that government had based their shielding letters on out of date databases. Hope that if further waves and lockdowns happen, the shielding letters will be sent out correctly.
- **Angela Burns MS:** Need to remember COVID is a respiratory disease and need to prioritise those and people who are immunosuppressed and at greater risk. Cannot put everyone into shielding - had to be cascade effect. Horrible if you feel like you should have been in it and weren't.
- **Graham Shortland:** One of the key issues - going back to specialist about need to shield. Written letters about returning to school/ employment.
- **Marie James:** Very little outcome data for rare disease patients who have had COVID-19 in general, this impacts on how people manage individual risk assessments. Son has Tuberculous Sclerosis and continue to shield as a family. Access to health care has been haphazard, routine appointments have been via teleconference which has been brilliant. Scans have been postponed which is a worry. Access to digital appointments fantastic.
- **Graham Shortland:** Individual societies are meeting around UK. COVID burden mild generally in children and no rise in admissions for metabolic disease in UK. Tapping into local specialists with that advice within disease cohorts. Generally children are doing very well and taking children off shielding list as this has become apparent through research.

## **5. Nick Meade (Genetic Alliance UK) - Launch of report, 'The Rare Reality of COVID-19'**

### **Some key challenges facing the rare disease community**

- Key workers worried about bringing COVID 19 back to shielding members of the family
- Parents who are carers for children becoming ill and unable to bring intensive support some children with rare conditions need. Extremely concerning.
- Family separation: One family separated for more than 6 weeks when child was already in a hospice. Various shielding reasons, had to stay separated.
- It's about how we have to take care to avoid COVID that is affecting people more than COVID 19 itself at the moment.

### **Launch of Report**

#### **Background**

- Launched on 2 July & article in Health Services Journal

- Letter sent to Secretary of State for Health
- Sources in report come from 2 areas - community check in meetings held with patient organisations members weekly throughout COVID 19 period. The rest of the information comes from a survey EURODIS ran early in COVID 19 period, UK data - 431 respondents, 65% patients and 29% carers

## **Key messages**

### **Interruption to Health Care**

- There's been a lot of interruption to health care. 1 in 5 thought interruption could be life threatening. Two thirds thought interruption was definitely detrimental to wellbeing.
- It will take a long time to find out about the true impact within the community due to the knock on effect throughout a persons life.
- Access to medicines was also affected with one fifth of population seeing an interruption.

### **Remote Healthcare**

- A lot of healthcare moved online during the outbreak. 50% of community had online consultation for the first time during the crisis. The experience was overall very positive. Small group who found it not at all useful. Cautious of being too positive towards telemedicine, always a group who can't access online consultations for accessibility reasons or because the healthcare they need requires face to face consultations. ``

### **Wider Impacts of COVID 19**

#### **• Communication around shielding**

These arrangements caused a great deal of worry and confusion in our community, for many reasons:

- Variation in guidance in the four nations of the UK, changing at different times, sometimes suddenly, without clear rationale.
  - The basis for the categorisation of conditions' and patients' risk levels has been slow to be explained.
  - Letters conferring the 'extremely clinically vulnerable' status came late, or not at all, and were allocated inconsistently.
  - The distinction between the two tiers of people told to protect themselves has led to those in the less protected tier feeling abandoned and isolated without support.
  - Many elements of real life have not been covered by the guidance – families with a child who must be shielded have not had clear guidance on what to do about a parent needing to go to work, or a sibling being invited back to school.
- Powerful to hear about how these problems impact on the rare reality of people affected by rare diseases live especially mental health and families.

## **Education and access to support**

Another aspect that has been making people's lives difficult, people have children who live with rare diseases especially those with Special Educational Needs and Disabilities (SEND) who rely on the education system to support them.

A series of issues have acted in parallel to amplify the impact on our community:

- The closure of schools has placed significant pressure on the parents of children with special educational needs and disabilities (SEND).

- Households are reluctant to admit local government provided carers because of the risk that Covid-19 presents.

- Reopening of schools is happening without due consideration to the needs of children with SEND, and in many cases without adequate staffing levels.

## **UK Strategy for Rare Diseases**

- Data and coordination of care elements of the 2013 UK Strategy for Rare Diseases would have undoubtedly helped had they been implemented properly:

- Telemedicine is discussed in the original strategy document as a tool for the delivery of care coordination

- Specific and comprehensive patient data for rare conditions would have facilitated the coordination of shielding information

## **Recommendations**

### **Continuing to deal with Covid-19**

- In monitoring the spread of Covid-19, data should be collected that will enable assessment of the impact, in terms of morbidity and mortality, on people living with rare conditions.

We haven't seen the collection of data but I am sure clinicians are collecting data and we will see some interesting research in coming months.

- People living with a rare condition who have a legitimate reason to ask for a test for Covid-19 (either for diagnosis or in order to assess future risk) should be given priority access.

- Access to PPE (personal protective equipment) should be guaranteed for people with rare conditions and those providing care and support to rare condition patients.

- When a vaccine or other prophylactic treatment becomes available, patients with rare conditions should be among those with priority access, provided their health condition allows.

## **Successful transition from crisis state**

-For those with rare conditions that confer high risk from Covid-19 who wish to continue protecting themselves through isolation, the support mechanism established should continue to be provided – no one should be forced to stop their protective isolation/ should be phased out gently

-A clear and short timetable for the reintroduction of services should be published to give people living with rare conditions clarity as to when they can expect a full service from the NHS.

-The provision of remote consultations should be continued. Care should be taken to integrate telemedicine into routine care practice with the necessary clinical assurance and data protection safeguards.

-Schools and educational facilities should be supported to ensure that they have the necessary flexibility to respond to individual needs and to adapt practices for children with rare conditions.

### **Learning for the future**

-The challenges and failures in delivering letters giving shielding advice should be examined, and actions should be taken following this analysis to ensure the UK is able to rapidly provide tailored public health advice to specific populations with specific conditions.

-The development of the new UK framework for rare diseases has been delayed by this crisis – it is now more necessary than ever. Learnings from this crisis should be incorporated into the new framework before the end of 2020.

### **6. Discussion after Nick's presentation:**

**Mike Cain:** consistency desired between home nations in support offered to patients with rare diseases

**Graham Shortland:** concerns raised with Welsh Health Specialised Services Committee (WHSSC) about access to cross border services impacted during pandemic.

**Marie James:** Concerns that tuberous sclerosis clinic will not be funded as resources are re-directed to COVID-19, patients accessing treatments at the moment through good will since 2015 rather than funded services. Concerns that access to treatment will become more difficult. Concern about losing clinical and research expertise. Prior to 2015, clinics were in Bath, Cambridge or St George's.

**ACTION: MB & EH - Letter to Vaughan Gething as soon as possible copied to Graham Shortland regarding TSC Clinic in Wales.**

**Graham Shortland:** Do have representation on UK Framework for Rare Diseases and want Wales to continue to be part of work for Framework development

**Tony Thorburn:** Support that report reflects views of rare disease community.

**Tony Crowhurst:** Throughout crisis we've heard about the number of excess deaths. We've not heard about quality of life issues or mental health issues/ loneliness and isolation comes up time and time again. This is something society has to deal with whether its a rare disease or disability generally.

**Angela Burns AM:** Really exercising us. Sat on Health and Social Care Committee and did a lot of work on this. With more lockdown coming, got to do more. Really difficult situation and I'm sure we can all relate to it. We are trying to work on it across all parties.

Clear list of people who have to shield/ those who would be extremely vulnerable on Welsh Government website.

**Tony Thornburn:** addressing Tony's point - only way to get a grip on this is a local response. Local Health authority get together with other agencies in town and working properly together. Difficulty at the moment is access to timely, informative local data to makes community based support more difficult. Local services and local people looking after each other is so important. Generate support from local services and local people looking after each other.

**Angela Burns MS:** Outstanding community initiatives and local councils have done a fantastic job locally. We have now got compassion fatigue/ crisis management fatigue following months, and coming back into another cycle of spiking and generating same community spirit isn't going to be as easy. Do take your point.

What would you like to end this session on? A report put to government with an overview of rare conditions/ what we want to see/ meeting to discuss a particular topic? Really lucky to have Graham Shortland - other groups we don't have access to people like Graham.

**Graham Shortland:** Useful if Graham/ Emma could have a session so Graham could ask members of group for 10-15 minutes to get feedback about direction.

2 side of A4 from Angela Burns MS as Chair would be powerful.

**Angela Burns MS:** Next meeting to be held in January 2020. Leave a legacy.

**Tony Crowhurst:** GPs should be more aware of some of the more common rare diseases and should be given training or information sheets be made more available to some GPs to make them more aware of the issues that are raised in groups like this.

**Graham Shortland:** Tried to look at education. Difficult to get a GP on the group. Hopeful of getting funding for a SWAN UK clinic. Myself and Sian Morgan from genetics talking to Paediatricians about WINGS programme - whole genome sequencing in sick neonates. Need to educate GPs and paediatricians. Something to go in the letter.

**Emma Hughes** - Rare Disease teaching weeks for health professionals – those interested in giving a session could be put in touch to raise awareness.

**Samuel Chawner:** Not just about awareness in GPs, awareness should be raised within Medical Genetics about mental health issues and vice versa mental health professionals should be made more aware of medical genetic issues. Connecting up areas is important.

**Tracy Blockwell:** Point made that in GOSH children have access to health psychologists, when they came back to Wales there was no equivalent. CAMHS and mental health services don't offer the same support if there's not a diagnosis of mental health issues leading to feelings of abandonment.

**Tony Thornburn:** Willingness of GPs/ secondary care clinicians to seek a second opinion and refer on is important. CONCORD - may want to emphasise this on the Welsh side.

**Angela Burns MS: Closes meeting**