

Behçet's UK 2020 Conference and AGM

The 2020 Behçet's UK Conference and Annual General Meeting took place online, supported by '1 Wimpole Street – home of The Royal Society of Medicine'. A total of 192 people registered to attend, of whom 133 attended live at some point during the conference. Of these, 106 were patients/carers and 109 lived in the UK. Feedback survey results indicate that 47% of attendees watched with two or more people.

Welcome and guide to the day



Michele Acton, CEO of the RSM, opened the meeting, highlighting the theme of 'new ways of working' and the vital importance of patient data. She noted the upcoming Behçet's programme in the RSM's 'Medicine and Me' series, which will bring together patients, clinicians and researchers. Tony Thornburn then welcomed everybody to the meeting and thanked the RSM for their support.

Importance of patient data

Building your patient data to drive research



Dr Rick Thompson is CEO of Findacure, a charity that aims to connect rare disease patient groups together so that they can learn from one another. He introduced a video made by **Daniel Lewi**, who started the CATS (Cure and Action for Tay–Sachs) Foundation in 2011 when his daughter was diagnosed with Tay–Sachs disease. At the time, there was no support available for parents of children with Tay–Sachs or the closely related Sandhoff disease, rare genetic disorders that reduce life expectancy to around 5 years. Because these disorders are very rare (1 in 320,000 people), it was necessary to form a European consortium to increase patient numbers and thus the perceived priority level.

Stakeholders (families, charities, researchers and pharmaceutical companies) have been brought together to collaborate in collecting data and investigating potential treatments. A registry was established, which now includes rich data from 120 patients, and research into the disorders and their treatment has greatly increased. Daniel stressed that data collection is hard work, and it took 5 years to collect enough

information (mostly patient-reported data) to be useful. Having a digital partner (OpenApp) speeded up the process, but **Con Hennessy**, MD of OpenApp, confirmed that a registry is a project that runs over decades rather than years. He agreed that a certain number of patients are needed to enable advocacy within health systems, and European networks are important for rare diseases. Daniel stressed that transparency about the reasons for data collection is key, and Con added that patient organisations/communities must own the data. He said that clinical data is needed as well as patient-reported data; tying clinical endpoints and quality of life data together can help to show the impact of the disease, and its treatment, on patients.

Rick concluded by saying that the example of the CATS Foundation showed what is possible in a rare disease if people work together.

Behçet's UK AGM



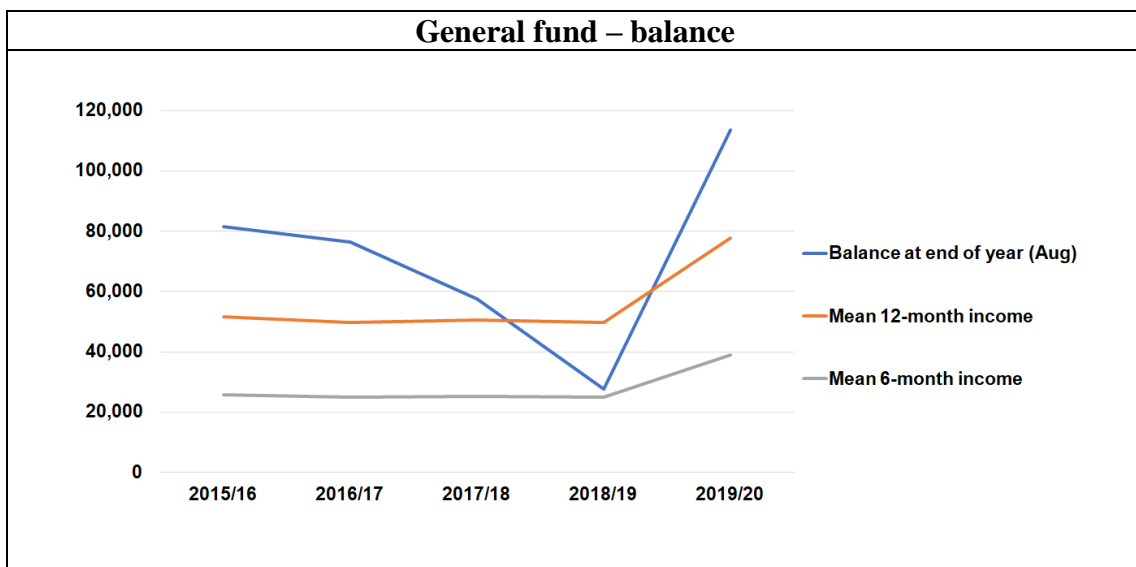
Tony Thornburn welcomed everybody to the AGM and encouraged them to read the Annual Report and Accounts, which are available on the website. He mentioned the stability in the list of Trustees over the past 3 years, noting that Richard West has stood down as Vice-Chair to take a well-earned break. Additional trustees offering specific skills or regional representation would be welcomed. The clarity of purpose and coherence of the charity has been maintained, and wider visibility has mostly been achieved this year through online attendance at meetings.

Tony thanked the 44 people who had returned the Priority Setting Partnership questionnaire, encouraging others to do the same. He also thanked the support group organisers who have recently stood down, as well as the ones who are continuing (Yvonne Morris, Fionnuala McKinley and Rachael Humphreys). He finished by thanking all the members who fundraise for Behçet's UK and acknowledging Deborah Cardinal's involvement in social media initiatives and Gemma Darlow's work to refresh the website.



Alan Lane reported that the Society now has 1134 full members, up from 1058 in 2019, with 21 junior members, 80 associates, 33 donors and 79 lapsed members. Patients account for 1084 members, with 52 supporters, 124 carers and 59 medical staff. Most (1093) members live in England, with 98 in Wales, 78 in Scotland, 36 in Northern Ireland and 7 in the Channel Islands. The proportion of members in receipt of state benefits or pensions is 81%; 74% of members are women, and most are in their 40s or 50s. Membership subscriptions remain at £20 per year for 2020/21. A typical grant award for members who meet the fund criteria is capped at £500, but in exceptional circumstances the maximum for any individual grant is £1000.

The Society's general fund went from being too high in 2016 to dangerously low in 2019 (the target is between 6 and 12 months' normal income). However, in March, a cheque for £100,000 was received from a member's estate following a 2-year long legal dispute. This is easily the largest donation in the charity's history, and the Trustees will use it to sustain the Society's activities over the next 3 years. Two-thirds of the 2019/20 income came from this legacy, with a further quarter coming from subscriptions and donations. Staff costs account for about 40% of expenditure. The research fund received a large donation in 2019, and the reserve is currently about £15,000. Alan noted that fundraising activities have increased this year and thanked everyone for their efforts.



Rachael Humphreys reminded the audience that Judith Buckle had started the Behçet's Syndrome Society (now Behçet's UK) in York in 1983. This year's Judith Buckle award went to Katy and Emma Davis, who died in April 2020. Rachael informed everyone how the twins had helped others through their work in nursing and their support to Behçet's UK members. They were regular visitors to Behçet's online forums and will be greatly missed by the community. The award was presented to their family (mother Julie, father Alan and sister Zoe) by the Mayor of Yeovil, and Rachael read out a letter from the family.



Improving patient outcomes

BAD/BSR guideline progress report

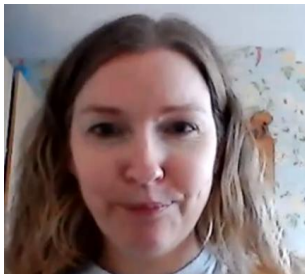
Dr Ruth Murphy, Consultant Dermatologist at Sheffield Teaching Hospitals NHS Foundation Trust, gave an update on the Behçet's guideline being developed jointly by the British Association of Dermatologists and British Society of Rheumatology. Its aims include driving greater understanding and increasing advocacy of the affected population, reflecting the patient journey and the range of disease presentations, and reflecting the need for multi-specialty and multi-professional input. The Guideline Development Group includes two patient representatives as well as a wide range of clinical specialists.

The guideline is being developed using GRADE (Grading of Recommendations Assessment, Development and Evaluation), which sets out criteria for quality of evidence and strength of recommendation. Evidence is categorised as high, moderate, low or very low quality. The process from evidence to recommendation is transparent,

and patient values and preferences are acknowledged. GRADE challenges guideline developers to specify all outcomes of importance to patients, and to differentiate outcomes that are critical for decision-making from those that are important but not critical and those that are less important. Recommendations are classified as strong or weak, with NICE stating that strongly recommended therapies should be offered to patients and weak ones considered.

The Behçet's Guideline Development Group has identified relevant randomised controlled trials for inclusion and has started to identify other (e.g. cohort, case-control) studies. The group has also started to write the narrative of the guideline.

Bike for Behçet's/Run for Rare 'Completion'



Rachael Humphreys reported on the success of a fundraising idea that she had during lockdown, which had helped with her own and others' mental health and raised valuable funds for Behçet's UK. Rachael had found early morning runs a great relief while she was shielding during lockdown, and her friend Laura had enjoyed cycling in the

quiet streets of London. The challenge to run or cycle the distance between the three centres of excellence – 226 miles – over 13 weeks was taken up by many people, some of whom 'met' other people with Behçet's for the first time through taking on the challenge.

People who took part included Clare Gibson, Lauren Bradfield, Stacey Rodger, Amanda Chapman, Laura and Zoe Forrest, and London Support Coordinator Jean Christians. John Mather cycled almost 1000 miles and also involved several of his friends, while the Skinner-Grant family raised over £4500 and Izzy Grant's school has nominated Behçet's UK as one of its charities for the year. An amazing total of £10,125 has been raised so far; Rachael thanked everyone involved and noted that the challenge can easily be replicated. All the stories can be read at <https://behcetsuk.org/news/bike-for-behcets-run-for-the-rare/>.

Assisting diagnosis – Diagnosing rare disease



Dr Will Evans, a GP and NIHR Practice Fellow at Nottingham University, spoke about using electronic health records (EHRs) to identify patients with rare diseases. He said that GPs are generalists, who offer family-centred care using the bio-psycho-social model with the consultation as the primary tool. There are 42,000 GPs in the UK seeing 1 million patients every day, which represents 90% of all patient contacts in the NHS and 9% of the NHS budget (or £136/patient/year). Collectively, rare diseases are not rare – in a typical GP practice of 8000 patients, 470 will have a rare disease. GPs diagnose the ‘bread and butter’ conditions, but they may be able to identify patients missed by secondary care by joining the dots between clinical features spread across organ systems, specialties and time.

It is now possible to enhance this process by using web-based resources such as Mendelapp to do a problem-based search for differential diagnoses. In future, clinical decision support systems embedded in EHRs will improve diagnosis. MendelScan is one such system. It uses pseudonymised data from GP networks and applies disease-specific algorithms to identify suspected cases of rare diseases using accepted clinical criteria, and then delivers a clinical report to the GP for each suspected case. The approach focuses on encoding accepted guidelines and diagnostic criteria and matching them to EHR coding norms.

A pilot in a population of 501,000 is currently returning reports in a 78,278 sub-population. Criteria for 70 diseases are used, including the International Criteria for Behçet’s Disease, with criteria being digitalised using codes for individual symptoms. An example patient diagnosed with Behçet’s at the age of 34 would have breached the algorithm at age 29. Retrospective analysis shows 51 cases of Behçet’s diagnosed in the 501,000 pilot population (a higher prevalence than expected). Of these, 14 (27%) could have been flagged 1–9 years earlier by MendelScan. The total number of cases flagged by the algorithm was 178. Internal review reduces the number by approximately 50% (based on experience of reviewing cases to date in the sub-population of 78,278). This reduces the total number of reports to be returned to approximately 82 (including the 14 diagnosed cases). A health economic assessment

suggests that earlier diagnosis of the 14 patients would have resulted in an average saving of £876 per patient in primary care costs. This work suggests that such an approach can work for Behçet's, with a probable acceptable sensitivity and specificity, and could lead to a cost saving.

Rudy Benfredj, CEO of Mendelian, commented that these results are very encouraging. The use of this type of approach is likely to increase, with eventual implementation at scale. The health economic results are important in enabling this to happen.

Centres of Excellence

Behçet's Patients Centres update



John Mather, BPC Operations Manager, began by saying that although COVID-19 has changed things dramatically, BPC has the same team as before and the same commitment to supporting patients. However, patients are now supported 'as and when' rather than at clinics, with no face-to-face contact. Flexible working includes expanded hours for the support coordinators, who try to contact every patient on the clinic list to offer support as necessary, as well as having regular catch-ups with the centre staff. They are also running a check-in service for patients known to be vulnerable, offering video call training to enable patients to maintain contact with their families, and working as NHS volunteer responders. In addition, BPC has been working alongside Behçet's UK to provide patients with non-medical COVID-19 support and information through <https://behcetpatients.org.uk/covid-19-information> and <https://behcetsuk.org/coronavirus-covid-19/>.

John finished by saying that more than 400 responses to the 2020 quality of life survey have been received, and these are being analysed by the London team. A COVID-19 patient survey has also been launched to discover the impact of the pandemic on patients' wellbeing and help with preparation for the winter months.

Patient data, new ways of working, quality of life headlines and medical panel updates



Dr Priyanka Chandratre, Consultant Rheumatologist in Birmingham, gave an update on the study investigating the epidemiology of Behçet's in the UK. A study in UK primary care in 2017 using The Health Improvement Network (THIN) reported a Behçet's prevalence of 14.6 per 100,000 person-years, much higher than previous estimates. Pilot work from the Birmingham Centre of Excellence aimed to calculate the prevalence and incidence of Behçet's using the adult population of the centre's catchment area (~16 million in 2015) as a denominator. Other objectives of the nationwide study are to collect descriptive statistics (demographics, clinical characteristics, end-organ damage, treatments and disease activity), analyse the association between independent variables and disease activity, and do cluster analysis of the clinical manifestations.

Of the 296 Behçet's patients included in the pilot, 80% had oral ulceration, 64% had genital ulceration, 51% had ocular manifestations and 32% were on biologic therapy. Genital ulceration and musculoskeletal problems were significantly more common in female patients, while ocular and neurological problems were more common in males; male patients also had higher disease activity scores. The work done in collaboration with primary care colleagues using THIN have recently been published in *Rheumatology* and show an increase in the prevalence of Behçet's over 17 years. This study also shows a higher risk of ischaemic heart disease, venous thrombosis and pulmonary embolism compared with controls.

The findings of the nationwide study will be validated by linking data from the centres of excellence to Hospital Episode Statistics data and the Clinical Practice Research Datalink. This will be the largest population-based study in the Western world and will give true estimates of prevalence and incidence, as well as greater understanding of disease/phenotype evolution which will allow informed policy making and resource allocation. The study has been match funded by Behçet's UK thanks to the generous sponsorship of Virginia Yee, with the other half coming from Sandwell and West Birmingham Hospitals NHS Trust. It will help to assess the

impact of the Behçet's centres and could serve as a starting point for a national disease registry.



Prof Rob Moots, Clinical Lead at the Liverpool Centre, spoke about remote consultations. He said that the three centres had remained open, but there had been fewer face-to-face consultations and more telephone and video consultations. For example, patients were being asked to send in photos of lesions for evaluation and storage on a secure server. Although he missed the informal chats with patients, and remote consultations are not ideal, Prof Moots said that he had learned a lot from working differently. Remote consultations will continue for some patients even when things are back to normal, as they suit some people better than coming in to the clinic. However, most patients would prefer to come in to the clinic, see the specialists in person and meet the support coordinator and other patients.

Dr Will Evans returned to give the GP perspective, saying that telephone and video consultations were being used as a first point of contact to triage patients, but face-to-face consultations should be available for those who need them. Telephone consultations were already well established in general practice, and video is now becoming routine. The pandemic has increased the speed of uptake of new technology, and it is very quick and easy to document skin lesions, for example, in electronic health records with the use of a smartphone.



Prof Farida Fortune, Clinical Lead at the London Centre, gave an update on the headlines from the quality of life survey. In the 2009 survey, mucocutaneous manifestations (oral/genital ulcers and skin lesions) were reported as the most important symptoms affecting quality of life, while joint problems and fatigue were more prominent in 2014. The findings in 2020 are similar to 2014, especially the prominence of fatigue. Prof Fortune suggested that the emphasis on oral health in recent years and the development of a bespoke mouthwash have had an impact, and now it is important to concentrate on joint health and management of fatigue. She said that a better diet has probably led to improved quality of life, and smoking has decreased greatly since

2014 with only 9% of patients now smoking. However, an increase in cannabis use could be having a negative effect, particularly in combination with smoking. Mental health is still an issue, with a big recent increase in anxiety.

Turning to the current situation, Prof Fortune said that many people with Behçet's had increased disease activity, which could be partly because stress is a known trigger. Some patients with active Behçet's had reported COVID-like symptoms (especially ocular and neurological symptoms), but none of them had tested positive. All patients should receive the flu vaccine, and pneumococcal vaccine would also be a good idea; the two vaccinations should be done separately and in different arms. Prof Fortune pointed out that the end of shielding was a difficult concept for some people. For those who have to return to work, employers have a responsibility to do a thorough risk assessment.

Medical Advisory Panel Q&A and Conference Close

The day finished with a question and answer session with members of the Medical Advisory Panel – Prof Rob Moots (Chair), Prof Farida Fortune, Prof Phil Murray, Dr Desmond Kidd and Dr Graham Wallace – as well Prof Dorian Haskard (President, ISBD), Dr Deva Situnayake (Clinical Lead, Birmingham), Dr Suzanne Crozier (Clinical Psychologist), Dr Priyanka Chandratre (Rheumatologist) and Dr Will Evans (GP and NIHR Practice Fellow). The panel answered many of the questions that had been submitted before and during the meeting.

Prof Murray emphasised the importance of patients developing eye problems being seen as a matter of urgency. Profs Moots and Fortune discussed fatigue as a key unmet need in Behçet's, which is more prominent now that better treatment has reduced the impact of other symptoms; the interrelationships between diet, sleep and fatigue need further investigation. The panel considered the possibility of patients using an app to record their symptoms on a daily basis – such patient-recorded outcomes are very important and could help to reveal patterns.

Clare Griffith, Editor