Disclosure of Interests: None declared

[Disclosure of Interests]

Nursing staff at Redcliffe Hospital and Administration officers at Redcliffe & North-West University of Queensland

Acknowledgements:

Scientific Abstracts


Disclosure of Interests: W. Gerhart1, M. T. Duruöz2, J. Lowe3, D. Webb4, L. Wermskog5, J. Davies6, R. Howard7, M. Mallinson8, C. L. Koehn9, Canadian Spondyloarthritis Association, Director, Toronto, Canada; 2Marmara University School of Medicine, PMR Department, Istanbul, Turkey; 3Axial spondyloarthritis International Federation, Projects, London, United Kingdom; 4National Axial Spondyloarthritis Society, Director, London, United Kingdom; 5Spafo Norge Spondyloarthritisforbundet, Director, Oslo, Norway; 6Axial spondyloarthritis International Federation, Executive, London, United Kingdom; 7Spondylitis Association of America, Executive, Los Angeles, United States of America; 8Axial spondyloarthritis International Federation, Volunteering, London, United Kingdom; 9Arthritis Consumer Experts, Director, Vancouver, Canada

Background: The current delay to diagnosis from symptom onset represents one of the greatest challenges in axial spondyloarthritis (axSpA). Research shows an average global delay of almost 7 years 1 – and as long as 15 years in some cases 2 – during which the condition can progress and cause irreversible damage. Data indicates that women wait longer than men for a diagnosis 2, and there has been very limited progress in reducing the time to axSpA diagnosis. The axSpA diagnosis delay has a hugely detrimental impact on an individual's quality of life. Because the disease frequently has early onset 3, individuals are left with untreated or incorrectly-treated symptoms at a formative period in their life course, whilst they await their diagnosis.

Objectives: The Axial Spondyloarthritis International Federation (ASIF) set out to coordinate a comprehensive evidence-based global review of the factors influencing the current axSpA diagnosis delay and to produce a definitive report that shines a light on these barriers, as well as providing a resource that can ultimately empower a range of international stakeholders to reduce this delay.

Methods: A full literature review was carried out to identify relevant available global evidence exploring the axSpA diagnosis delay. In autumn 2020 ASIF held two virtual global forum events, involving patients and patient group representatives, researchers, rheumatologists and other healthcare professionals, to methodically explore key diagnosis challenges across different healthcare systems and the opportunities for addressing these. Break-out discussions were held and participants were asked to identify the personal and societal effects of the diagnostic delay; the reasons it occurs; and initiatives to tackle the challenge. Alongside key stakeholder testimonies, best practices from around the world were also identified. 92 stakeholders participated in the events; they represented patients and healthcare professionals from 23 countries across five continents.

Results: The findings from these activities were incorporated within a new 'Delay to Diagnosis' report, which for the first time definitively sets out the lived reality from a global perspective of the axSpA diagnosis delay. The report identified important commonalities across different countries and healthcare systems contributing to the current average global 7-year diagnosis delay, including:

- Poor awareness of axSpA, particularly in primary care services
- Complexities in diagnosing the disease
- Poorly defined referral pathways
- Insufficient patient access to rheumatologists and appropriate diagnostics

The report also highlights the significant impact this delay has on individuals and wider society, providing a foundation for future advocacy work. A series of recommendations have also been identified, the implementation of which will help to instigate tangible progress in reducing the current delay.

Conclusion: Despite longstanding challenges, there are now clear opportunities for transforming how axSpA is diagnosed around the world. This message needs to be heard and acted upon urgently by all those involved in the management and delivery of axSpA care. The future programme of work for ASIF's Delay to Diagnosis project will respond to these findings and be centred around supporting axSpA patient associations globally to take this call to action forward throughout 2021 and beyond.

References:

1. Zhao et al; Diagnostic delay in axial spondyloarthritis: a systematic review and meta-analysis; Rheumatology, 2021
4. Disclosure of Interests: Wendy Gerhart Employee of: I was employed by Janssen Canada from 1992 - 2017, Mehmet Tuncay Duruöz: None declared, Jo Lowe Grant/research support from: No financial grants received individually; however, my role as Project Manager at ASIF is currently funded partially, by UCB Global and partially by Novartis Global, Dale Webb: Speaker bureau: Janssen and Novartis, Grant/research support from: Individually, no. But NASS receives grants from Abbvie, Biogen, Eli Lilly, Janssen, Novartis and UCB, Lil-lann Wermiskog Grant/research support from: Individually, no. But Spafo Norge receives a small amount of funding for ongoing projects from Novartis... Jo Davies Grant/research support from: Individually, no. However, ASIF are currently funded by UCB, Lilly, Boehringer Ingelheim, Janssen and Novartis; this funding partially covers staff salaries as well as a range of projects, the content and outputs of which are not influenced by the funders... Richard Howard Shareholder of: Abbvie, Amgen, Bristol-Myers Squibb, GSK, Johnson & Johnson, Eli Lilly, Merck, Novartis, Pfizer, and Teva. I own <20 shares of any one stock and these stocks represent <4% of personal investments, Consultant of: Yes, GSK, Novartis - but then donated to Spondylitis Association of America, Grant/research support from: I have not personally received financial grants. SAA has received financial support from Abbvie, Amgen, BI, J&J, Lilly, Novartis, Pfizer, UCB; R. Howard Consultant of: Abbvie, Amgen, Eli Lilly, Janssen, Merck, Pipeline, and UCB; Michael Mallinson Consultant of: No. But, for full disclosure: I have received honoraria in the past, for participating in patient advisory board activities, from Abbvie, Novartis, Pfizer and UCB; Cheryl L Koehn Grant/research support from: OUR ORGANIZATION, ARTHRITIS CONSUMER EXPERTS, HAS. I HAVE NOT AS AN INDIVIDUAL. HERE IS OUR PUBLICLY AVAILABLE DISCLOSURE ON ALL ONLINE AND PRINT MATERIALS, PRESENTATIONS, MEETINGS, GOVERNMENT CONSULTATIONS: https://jointhealth.org/about-principles.cfm?locale=en-CA
5. Over the past 12 months, ACE received grants-in-aid from: Arthritis Research Canada, Amgen Canada, Canadian Institutes of Health Research, Canadian Rheumatology Association, Eli Lilly Canada, Hoffman-La Roche Canada Ltd., Knowledge Translation Canada, Merck Canada, Novartis Canada, Pfizer Canada, Sandoz Canada, Sanofi Canada, St. Paul’s Hospital (Vancouver), UCB Canada, and the University of British Columbia.

Background: Research carried out in 2016 by NASS showed that the range and quality of axial spondyloarthritis (axSpA) services generally offered around the UK were variable 1. The publication by the regulator, the National Institute for Health and Care Excellence (NICE), of a Guideline for Spondyloarthritis (NG65) in 2017 2 and the corresponding Quality Standard (QS170) in 2018 3, for the first time provided national guidance and standards of services that should be available for people with axSpA. NASS national oversight of the implementation of these recommendations was missing.

Objectives: NASS worked with Parliamentarians to establish the All-Party Parliamentary Group for Axial Spondyloarthritis in January 2019. We gave it a very specific objective - to oversee the implementation of NHSE and QS170. The group seeks to improve axial SpA services in England whilst raising awareness of the condition at a parliamentary level, working closely with NASS.

Methods: The group is a unique forum in the UK, bringing together patients, clinicians, researchers, policy makers, national bodies and parliamentarians. The group has met five times covering a range of topics including the delay to diagnosis, the uptake of NG65 and hydrotherapy. In 2019 the group carried out a national inquiry into the standards of axSpA services in the UK, developing a ten-question quality framework, based largely on the NICE Guideline recommendations and Quality Standard. In July 2020 a meeting was convened to discuss the impact of COVID-19 on axSpA services. The meeting presented research carried out by NASS and M&F Health Communications on September 16, 2023 by guest. Protected by copyright.http://ard.bmj.com/ Ann Rheum Dis: first published as 10.1136/annrheumdis-2021-eular.2612 on 19 May 2021. Downloaded from http://ard.bmj.com/ on 19 May 2021. Copyright © 2021 BMJ Publishing Group Ltd. All rights reserved.


Results: The results of the national Inquiry for England were published in January 2021 4. The disparity in the level of services remains in the provision of axSpA services. Only 21% of local commissioning bodies have an inflammatory back pain pathway, and less than half of NHS providers have a specialist axial SpA clinic. The results of COVID survey shows significant impacts on the health of axial SpA patients and on the availability and modality of rheumatology services. The results of the above publish showed that with a set of recommendations, creating minimum service specifications for axSpA services during crisis periods such as the recent pandemic, as well as service recovery. Comment on this research was also published in The Lancet Rheumatology 5.
In September 2020 the work of the APPG led to a debate in Parliament on delayed diagnosis in axial SpA.

Discussions on the future of hydrotherapy services has resulted in the mobilisation of stakeholders across condition areas.

Conclusion: All Party Parliamentary Group is already proving to be an effective political lever to improve axial SpA care. It has shown huge variations in the standard of care and provision of services still remain and has brought this to the attention of Parliamentarians, policy makers and clinicians.

REFERENCES:
[1] Mohammad H Derakshani, Himanshu Pathak, Debbie Cook, Sandy Dickson, Stefan Siebert, Karl Gaffney, NASS and BRITSpA investigators, Services for spondyloarthritis: a survey of patients and rheumatologists, Rheumatology, February 2018

Disclosure of Interests: All Jill Hamilton Grant/research support from: Funding was received from Novartis to support APPG 1, 2 and 4. Dale Webb Grant/research support from: Funding was received from Novartis for APPG 1, 2 and 4. Simon Whalley: None declared
DOI: 10.1136/annrheumdis-2021-eular.2612

OP027-PARE

METAVERN, THE EUROPEAN REFERENCE NETWORK FOR RARE HEREDITARY DISEASES: STRUCTURE, OBJECTIVES, METABERN RMDS AND THE ROLES OF EUROPEAN PATIENT ADVOCACY GROUPS (EPAGS)

L. Wagner1, S. Sestini2, C. Brown3, A. Finglas4, R. Francisco5, S. Bond6, C. Lampe7, C. Belettato6, C. Van Lingen6, M. Scarpa6, on behalf of MetabERN collaboration group.1Deutscheschwarz die Selbsthilfegruppe für Akzeptonure, e.V., DSAKU e.V., Stuttgart, Germany; 2Associazione italiana dei malati di acaptonuria, AIMAKU Siena, Italy; 3Krabbke UK, Krabbke UK, London United Kingdom; 4MDS Action Foundation, MDS Action Foundation, Dublin, Ireland; 5Portuguese Association for CDG and Other Rare Metabolic Diseases, APCDG-DMR, Almada, Portugal; 6Udine University Hospital, MetabERN, Regional Coordination Center for Rare Diseases, Udine, Italy; 7Center for Rare Diseases Giessen, ZSEGI, Giessen, Germany

Background: Inborn metabolic disorders (IMDs) currently encompass more than 1,500 diseases with new ones still to be identified1. Each of them is characterised by a genetic defect affecting a metabolic pathway. Only few of them have curative treatments, that target the respective metabolic pathway. Commonly, treatment examples include diet, substrate reduction therapies, enzyme replacement therapies, gene therapy and biologicals, enabling IMD-patient now to survive to adulthood. About 30 % of all IMDs involve the musculoskeletal system and are here referred to as rare metabolic RMDs. Generally, IMDs are very heterogenous with respect to symptoms and severity, often being systemic and affecting more children than adults. Thus, challenges include certified advanced training of adult metabolic experts, standardisation training plans, social support and development of therapies for diseases that do not have any cure yet.

Objectives: Introduction of MetabERN, its structure and objectives, highlighting on the unique features and challenges of metabolic RMDs and describing the involvement of patient representation in MetabERN.

Methods: MetabERN is stratified in 7 subnetworks (SNW) according to the respective metabolic pathways and 9 work packages (WP), including administration, dissemination, guidelines, virtual counselling framework, research/ clinical trials, contempt of care, education and patient involvement. The patient board involves a steering committee and single point of contacts for each subnetwork and work package, respectively2. Projects include identifying the need of implementing social science to assess the psycho-socio-economic burden of IMDs, webinars on IMDs and their transition as well as surveys on the impact of COVID-19 on IMD-patients and health care providers (HCPs), social assistance for IMD-patients and analysing the transition landscape within Europe.

Results: The MetabERN structure enables bundling of expertise, capacity building and knowledge transfer for faster diagnosis and better health care. Rare metabolic RMDs are present in all SNWs that require unique treatments according to their metabolic pathways. Implementation of social science to assess the psycho-socio-economic burden of IMDs is still underused. Involvement of patient representatives is essential for a holistic healthcare not only focusing on clinical care, but also on the quality of life for IMD-patients. Surveys identified unmet needs of patient care, patients having little information on national support systems and structural deficits of healthcare systems to ensure HCP can provide adequate clinical care during transition phases. These results are collected by MetabERN and forwarded to the Directorate-General for Health and Food Safety (DG SANTE) of the European Commission (EC) to be addressed further.

Conclusion: MetabERN offers an infrastructure of virtual healthcare for patients with IMDs. Thus, in collaboration with ERN ReCONNET, MetabERN can assist in identifying rare metabolic disorders of RMDs to shorten the odyssey of diagnosis and advise on their respective therapies. On the other hand, MetabERN can benefit from EU-LAR’s longstanding experience regarding issues affecting the quality of life, all RMD patients are facing, such as pain, stiffness, fatigue, rehabilitation, maintaining work and disability claims.

REFERENCES:

S. Chibnall-Smith1, A. L. Chibnall-Smith1, R. Beesley1. 1Juvenile Arthritis Research, JAR Project, Tonbridge, United Kingdom

Acknowledgements: The authors thank the MetabERN collaboration group, the single point of contacts (SPOC) of the MetabERN patient board and the Transition Project Working Group (TPWG)

Disclosure of Interests: None declared
DOI: 10.1136/annrheumdis-2021-eular.3372

OP028-PARE

PROVIDING SUPPORT TO FAMILIES OF CHILDREN NEWLY-DIAGNOSED WITH CHILDHOOD ARTHRITIS: A PATIENT AND PARENT-LED PILOT STUDY TO DEVELOP AND ASSESS A LITTLE BOX OF HOPE® SUPPORT PACKS

S. Chibnall-Smith1, A. L. Chibnall-Smith1, R. Beesley1. 1Juvenile Arthritis Research, JAR Project, Tonbridge, United Kingdom

Background: Juvenile Idiopathic Arthritis (JIA) is a heterogeneous group of autoimmune disorders characterised by chronic joint inflammation, diagnosed in around 1 in 1,000 children and young people (CYP) under the age of 16. Delays in diagnosis are common [1], awareness is low, and paediatric rheumatological conditions have a considerable impact on young people and their families [2]. A lack of understanding amongst families of newly-diagnosed children leads to uncertainty and anxiety.

Objectives: This patient and parent-led project developed a resource pack for families newly diagnosed with JIA, to provide information and support. Following a pilot, feedback from recipients was collated and analysed to help improve future provision.

Methods: A young person with JIA identified the need for direct family support. Juvenile Arthritis Research (a UK charity) developed a unique pack of support information, containing resources for both children and their families - called A Horsebox Of Hope. This included information about JIA and support services available for families, as well as Kipo (a children’s book about JIA) and accompanying finger puppet. Clinicians at one paediatric rheumatology centre provided information about the packs to newly diagnosed families, who then requested a free box to be posted to them.