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MEETING ABSTRACTS

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Poster Presentations

P1

The first effective treatment for alkaptonuria; a collaborative, patient centric effort

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In 1902, Sir Archibald Garrod described alkaptonuria (AKU) as inherited for the first time. Since then, there has been little or no research into a treatment for the life-changing disease that affects 1 in 250,000 people worldwide. The AKU Society was founded in 2003 to find a treatment.
In 2012 the AKU Society founded a pan-European consortium, called DevelopAKUre. This was made up of 12 members, including hospitals, pharmaceutical companies, universities, biotechs and national AKU patient groups from all over Europe who shared one aim: to prove that a repurposed drug called nitisinone works in reducing the chemical that causes the damage of AKU and that it has a positive impact on clinical features.
Together, DevelopAKUre applied for funding through the European Commission's Seventh Framework Programme (FP7) in order to develop and run the trials that would prove the drug works. The funding secured for this programme included €6 million from the European Commission, with an additional €4 million in co-financing (for in-kind costs such as the drug supply)
Armed with this funding, we launched three studies into the efficacy and safety of the drug. The last, SONIA 2 (Suitability of Nitisinone in Alkaptonuria 2) was designed to answer once and for all if nitisinone can be used in AKU to reduce the chemical that causes the damage to bones and cartilage. The trial finished in January 2019. We found out at the end of 2019 that we had been successful. Sobi, the company that owns the rights to nitisinone, has now applied for a license for the drug's use to treat AKU across Europe.
This is the first time an effective treatment for AKU has been found. Nitisinone has now been proven to reduce the chemical that causes AKU by up to 99%. If given early enough, the drug could prevent the disease features from developing at all.

P2

'Patient journeys': Personal experiences shaping clinical priorities

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European Reference Networks (ERNs) are networks of highly specialised healthcare providers from across Europe. There are 24 networks, each focusing on a different rare disease area. ERN CRANIO is the ERN for rare and/or complex craniofacial anomalies and ear, nose and throat disorders. ERN CRANIO pools together the available disease-specific expertise, knowledge and resources from across Europe and initiates network activities designed to support patients, families and clinicians. The network also involves patient representatives in this action, who are able to engage with their patient communities and voice the needs and perspectives of patients and families.
Patient representatives within ERN CRANIO have reflected on their own personal experiences and engaged with their patient communities to develop patient journeys (PJs) [1]. PJs are disease-specific visuals, mapping identified patient/family 'needs' and 'ideal support scenarios' at key clinical stages. PJs have been developed for: Apert Syndrome, Genetic Hearing Loss, Pierre Robin Sequence, Craniofacial Microsomia, Cleft Lip/Palate and a PJ for Treacher Collins Syndrome is underway. These visuals can be continually shared, added to and developed.
The PJs can play a part in validating existing disease-specific network activities and guiding future focus. On initial inspection it also appears there is a common need for clear, easy to understand, medically accurate information on clinical diagnosis and/or treatment options. This serves to validate ERN CRANIO's strategic focus on the development of clinical guidelines. It also highlights the importance of ensuring these guidelines are made accessible to patients and families and are communicated clearly. This may involve creating a patient version of a medical guideline, for example.
A more in-depth inspection and systematic assessment of the common needs will need to follow. However, this initial finding highlights the potential value of using the PJs to validate the network's strategic focus and shape priorities on a broader level.

Acknowledgements: ERN CRANIO patient representatives have facilitated/are facilitating the completion of the patient journey content (Gareth Davies, Michel Francois, Jeroen van de Koppel, Thomas Luck,

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Quality assurance of rare disease (RD) Centers of Excellence (CoE) through designation, accreditation, monitoring and constant improvement provides a means to ensure high quality, centralization of resources and expertise, and cost-efficiency. EUCERD recommendations for quality criteria of CoE, issued in 2011, are still highly relevant [1]. In the State of Art Resource, almost all European Union (EU) Member States (MS) claim, that their CoEs conform to EUCERD recommendations [2]. However, national quality assurance processes differ significantly: some MS apply robust procedures, while in other MS, many of them - but not exclusively - are EU-13 MS, processes of quality assurance are less developed. Under the subsidiarity principle embedded into European treaties, the EU plays a limited role in many areas of healthcare, and CoEs quality assurance processes are a choice and responsibility of MS.
With the establishment of ERNs, another layer of quality assurance has been developed by the European Commission and the MS [3]. This new quality assurance framework may be in line, or not, with national accreditation systems and involves i) assessment of CoEs when they apply for Full Membership of ERNs and ii) continuous monitoring afterwards [4]. In every ERN, Members have to be "equal partners in the game" and share the same goals, rights and obligations. While the ERN logo should eventually be a quality mark of the highest standards, strong links of ERN Members to national systems, including many more and less specialized healthcare providers, are essential to ensure proper care pathways for RD patients.
Importantly, ERNs themselves and patients/non-governmental organizations provide us with additional means of "informal quality assurance". Many ERNs are implementing their own monitoring processes through the creation of registries to collect health outcomes that allow peer-benchmarking. Meanwhile, patients provide their strong voice through European Patient Advocacy Groups (ePAGs) and help to signpost "the best" CoEs through information sharing. In both these processes, the power of open, transparent information on performance may finally lead to improved transparency and accountability at a national level and, presumably, may have an impact on the composition of ERNs in the future.

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Patient's view on disruptive innovations in clinical research

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In order to improve clinical research, patient preferences and outcome measures relevant to patients should become the core of drug development and be implemented from the earliest stage of drug development. From 'bedside to bench' instead of from 'bench to bedside'.
At all levels the reuse of data could and should be enhanced. Patient derived or provided data are not owned by those who collected them, and their reuse should be primarily controlled by the donors of these data. Researchers and Health professionals are custodians (GDPR). To enable the optimal reuse of real world data, the data needs to be Findable, Accessible, Interoperable and Reusable (FAIR) by medical professionals, patients and in particular also by machines. For this reason the World Duchenne Organization published a Duchenne FAIR Data Declaration [1]. Reuse of placebo data and use of natural history data could speed up research especially in the field of Rare Diseases
At this moment, in line with GDPR, patients are in a good position to decide about the reuse of their own data and should not only have access to these data but preferable also be in charge of their own data. By collecting relevant data (Patient Reported Outcome Measures), storing their own clinical data from different data silo's (Electronic Health Records (EHRs), registries and companies) and deciding themselves with whom to share. Duchenne Parent Project together with Foundation29 set up a Duchenne Data Platform that facilitates all mentioned before.
Also trial design could profit from the reuse of data. Input from patients in trial design in general should improve as well. Projects such as trials@home (IMI) may help to change the future of clinical trials and the use of Community Advisory Boards (EURORDIS) should be encouraged as much as possible.

Reference

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S22

Repurposing technology to create a new normal for hidden disabilities

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Background: Drug repurposing for rare disease has brought more cost-effective and timely treatment options to patients compared to traditional orphan drug development, however this approach focuses purely on medical interventions and requires extensive clinical trials prior to approval. In the case of refractory epilepsy, practical solutions are also required to better manage daily life. Here we present an example of technology repurposing as a practical aid to managing absence epilepsy.
Methodology: Existing research tells us that seizure control is not the only consideration of Quality of Life in children with epilepsy and that mental health and caregiver/peer support are of utmost importance. We explored the needs of stakeholders and determined that there was a delicate balance between the individual (and those that care for them) and those that have the power to change their lives.
Results: Across all stakeholders there was a shared common need to obtain objective data on absence monitoring to relieve the burden on families/carers to retain manual seizure diaries whilst providing accurate and timely data to medical teams, researchers and social care.
Epihunter is an absence seizure tracking software using repurposed technology: a headset from wellness/leisure to collect electroencephalographic (EEG) data and an AI algorithm to detect and record absence seizures on a mobile phone application in real-time. Both EEG and video recording of the seizure are automatically captured. This low cost, easy

to use technology has been used in the home to provide objective seizure data prior to upcoming clinic appointments. The COVID-19 pandemic has prompted an acceleration in telemedicine and epihunter has improved the effectiveness of virtual consultations bringing opportunities for both diagnostics and informed changes in treatment.
Conclusions: Epihunter is an example of technology repurposing to create a new normal for people with hidden disabilities such as those living with absence epilepsy.

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Patients Perspective on 'need for centralization of care' and mobilizing system change

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2013 in Germany the National Action Plan for Rare Diseases (NAMSE) has been published with a detailed description of criteria on three levels:

1. A-Centers include research and training activities, are an expert center for more than one diagnosis.
2. B-Centers fulfill all criteria for one diagnosis and are collaborating with an A-Center and other B-Centers for the same diagnosis.
3. C-Centers are working together with a B-Center for the local/regional care of the patients.

The rare disease patient community tried to get this well detailed plan to be transferred to regulation which usually means an adequate financial substitution of those expert services.
The patients should benefit from a centralized expert treatment/care pathway.
Esophageal atresia (EA) is a rare congenital condition with an estimated prevalence of 1 to 2 in 5,000 live births. Esophageal atresia patients require life-long attention.
ERNICA has developed a 'patient journey' for EA patients, under the leadership of patient representatives from the international federation of EA support groups (EAT).

Fig. 1 Example of patient journey stage depicted pictorially

In Germany, patients with congenital malformations which need surgery in early life are treated in hospitals with (very) low experience.
How can we as patient representatives get the fruits of the ERNs into the national health system?
We don't have public money. We have no official contract and no political support.
KEKS e.V., the German EA support group together with other support groups (e.g. SoMA e.V.), and with surgical expert teams across Germany, some of them members in ERNs, started to organize monthly virtual boards for those patients.
A self-commitment on ethical and medical standards following the ERN-criteria, and a collaborative attitude within the group, help us to get step by step the first ERNICA results to the bedside of EA patients.

Publisher's Note