FESTIVAL OF GENOMICS LONDON

#GENOMICSFEST

Panel Discussions

led by key industry speakers

LIVE LOUNGE

"For patients, with patients: learning from patient experiences to maximise the benefit of clinical genomics"

Day 2 : 31st January 2018

Time: 13:20 - 14:15




Chairperson

Beverly Searle, CEO, **Unique – Understanding Chromosome & Gene Disorders**

Speakers

Arti Patel, Information Officer, **Unique – Understanding Chromosome & Gene Disorders**

Mariana Campos, Membership and Public Engagement Manager, **Genetic Alliance UK**

Jillian Hastings Ward, Chair of the Participant Panel, 100,000 Genomes Project, **Genomics England**

The Diagnostic Odyssey

- The time taken between a patient first developing symptoms of their condition and receiving a medical diagnosis.
- Patients face significant delays on their journey to secure a diagnosis.
- The majority of patients receive at least one incorrect diagnosis/diagnoses, and visit numerous doctors, before they receive a final diagnosis.
- On average, rare disease patients consult with 5 doctors, receive 3 misdiagnoses and wait 4 years before receiving a final diagnosis.
- Patients can experience issues in persuading medical professionals to believe their symptoms and describe how their condition is initially written off as ‘psychological’ or, parents are described as ‘neurotic’.

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

A Personal Experience from Jillian Hastings Ward

- Enrolled son in 100k Genomes project in Sept 15. Recruited as a trio: mum / dad / child (18 months).
- Consented to project capturing our Whole Genome Sequences and longitudinal health record (birth to death, eventually) and storing for future research.
- Sounds quite intimidating, and people who have more choice whether or not to join project may have been put off by this.
- But it was our best chance of finding answers so we agreed.
- The 100k Project set up, with actual participants overseeing what happens to the data, made it easier for us to TRUST that Genomics England will use it wisely.

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Looking for Support – SWAN

- It is estimated that around 6,000 children are born every year with a syndrome without a name . Why?
  - The condition is the rarest of the rare – never been seen before so not tested for.
  - Unusual presentation of known condition – clinical signs don't trigger testing.
  - Genetic changes found are of unknown clinical significance
- 50% of children undergoing genetic testing in the UK won't get a confirmed diagnosis.
- 75%-80% taking part in research studies (DDD, 100,000 Genomes Project)
- Graduate SWANs – received diagnosis but many continue to feel isolation - follow up support via SWAN UK, Unique or help to create their support group
- Support via a group appropriate to the diagnosis

www.undiagnosed.org.uk

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Receiving the Diagnosis

- Imagine you've been waiting 3 years to hear what's wrong with your son. Then imagine the phone rings out of the blue, and it's a doctor you've met once rushing to tell you some monumental news without first giving you a chance to catch your breath / finish paying for your shopping / swallow your lunch…
- Thankfully we had some good news, and it was delivered very well.
- Consultant (geneticist) emailed on a Monday afternoon to say she had something she wanted to discuss with us, and asked us to pick a time (we chose the next morning). Time to think but not too much time.
- Patient directly involved in choosing when they hear. Accessible professional, available for follow-up questions.
- No surprise letter at breakfast. No long wait for a clinic appointment, knowing some news is coming. No anonymous, inaccessible professional to deliver the news and walk away.

Delivering the Diagnosis

- Unique Paper on survey of experiences of diagnosis and genetic counselling of UK Families with a rare chromosome disorder
- Quantitative paper published in Clinical Genetics based on the experiences of 1,158 families replies
- Diagnoses were made by G-banding or array CGH analyses
- 7 stages of the patient journey were examined:
  - (i) Pre-testing process (ii) Communication of test result (iii) Referral to genetics specialist) (iv) Conduct of genetic consultation (v) provision of genetic & clinical information (vi) genetics service follow up (vii) sign posting to peer support group
- Little change in the experiences of many families from when diagnoses were made by g-banding or microarray analysis in the Unique paper when compared to the report produced by GA in 2016 about the impact of NGS
- In the Unique paper, 28 aspects of patient journey were recognised as requiring improvement; of these, only 12/28 are currently incorporated in NHS England's service specifications
- Funding permitted, Unique will repeat survey in 2020.
- doi:10.1111/cge.13207

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Undiagnosed Genetic conditions & the impact of genome sequencing

Report run by Rare Disease UK (https://www.raredisease.org.uk/our-work/undiagnosed-genetic-conditions-and-the-impact-of-genome-sequencing-2016/)

Main findings:

- Managing expectations of families including that:
  - the likely result would not be straightforward but very likely a series of letter and numbers relating to a gene change;
  - the results wouldn't lead to a prognosis or provide information about the future for their child
- Support for diagnosed:
  - Receiving a diagnosis has a huge emotional impact and the support families receive does not appear to be consistent
  - Families value being offered support from professional including genetic counsellors, but there appears to be inconsistency around what is offered.
- Post diagnosis care:
  - Patient organisations play an important role in providing families with a safe space to connect and come to terms with it.

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Impact of receiving a diagnosis

- Initial feeling was like completing a jigsaw but finding that the picture has a huge misty hole in the middle.
- But, at least we knew it was de novo, putting rest of family at ease.
- As time passes, it's more like being given a flag, that you can finally take to a hilltop and wave and see who else is waving back.
- 21st Century means we can find people all over the world within hours; amazing!
- Treatment may not be invented yet, but at least we can find others with same condition and compare notes….

Looking for Support – Unique

- Supporting families with rare chromosome and (autosomal dominant) single gene disorders associated with development delay and/or intellectual disability.
    - Provide information and support about their diagnosis
    - Support from other families with similar symptoms/challenges
    - Telephone and email helplines, social media support groups
    - Unique Database to match up families – not alone
    - Conferences & family days for families & professionals to meet others
    - Practical guides to help with daily living
    - www.rarechromo.org

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Building Rare Communities Project

- For disorder-specific groups not covered by Unique and others
- For families who have a diagnosis but no support group to support them
- They can be part of a condition specific network but with the support of a wider organisation
- Meet other patients/parents of children affected by the same condition, share experiences, face isolation
- Building Rare Communities – help patients/parents identify the needs of the community. Support with the day to day activities of setting up a group, being part of a community of other patients/parents setting up their group.
- Avoid the isolation of a patient group/fragmentation of the patient group community.

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

The Unique Registry

- A patient-based database to collate lifetime natural histories of the rare genomic disorders - information contributed by families themselves
- It's not just about clinical diagnoses, but also includes social, educational, developmental, behavioural and health information. Useful therapies and support from other sources too.
- Unique provides anonymised phenotype reports for professionals to help with counselling. Genotype database available on Unique website.
- Unique acts as a conduit between researchers and families for involvement in research projects
- Unique uses information from our database & from the medical literature to develop our peer reviewed disorder-specific information guides and leaflets, many now about single gene disorders as well as RCDs and many translated into numerous other languages– used by both families and professionals

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Challenges & hopes for the diagnosis

- Hope for improved clinical applications/care management
- Treating the needs/symptoms of the patient – not just the diagnosis
- Potential for treatments/therapies/ even a cure?
- Collating the information to find out what works best for patients with a particular disorder and sharing this information!
- Psychological impact of the diagnosis – “what’s the use if it doesn’t change anything” (+ve to have an answer!)/unfulfillment of and management of patients’ expectations/ ”not a bad parent”/validation of concerns/not the only ones/closure/hope for the future
- Families desire for research – both pure and applied

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

Let’s get thinking!

- How do you involve patients in your work?
- If you were offered WGS, in search of a diagnosis for your own condition or that of a loved one, would you take it?
- What kinds of questions would you ask before agreeing to give your sample?
- If you were a patient, what would you expect/hope to gain from receiving a diagnosis?

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

References

- Unique Paper

Szczepura, A., Wynn, S., Searle, B., Khan, A.J., Palmer, T., Biggerstaff, D., Elliott, J. and Hultén, M.A., UK Families with Children with Rare Chromosome Disorders: Changing Experiences of Diagnosis and Counselling (2003 to 2013). *Clinical Genetics*. Accepted Author Manuscript. doi:10.1111/cge.13207 http://onlinelibrary.wiley.com/doi/10.1111/cge.13207/full

- The Rare Reality

https://www.raredisease.org.uk/our-work/the-rare-reality-an-insight-into-the-patient-and-family-experience-of-rare-disease-2016

- Undiagnosed Genetic Conditions and the Impact of Genomic Sequencing (2016)

https://www.raredisease.org.uk/our-work/undiagnosed-genetic-conditions-and-the-impact-of-genome-sequencing-2016/

Understanding chromosome disorders Unique

Genomics england

GENETIC ALLIANCE UK

With special thanks to Jillian Hastings Ward

Understanding chromosome disorders Unique

www.rarechromo.org

Genomics england

www.genomicsengland.co.uk

GENETIC ALLIANCE UK

www.geneticalliance.org.uk