Metabolic Support UK are proud to present 'Rare Thinking', a three day festival of ideas, connections and opportunities for people living with Inherited Metabolic Disorders (IMD's) and their communities.

The Covid-19 pandemic has changed the landscape for rare disease patients, from the use of telemedicine to a reframing of quality of life and wellbeing. Amidst these changes, the UK rare disease landscape is undergoing increasing change through consultations on topics such as technology appraisal processes and regulation and the Rare Disease Framework. Alongside these wider movements in our sector, all impacting the diagnostic odyssey and equity of treatments, we have also seen shifts in the lived experiences of our patients.

Our festival is an exploration, asking how we respond to a changing landscape to ensure our rare communities are not left behind.

'LET'S CHANGE THE CONVERSATION FOR PEOPLE WITH INHERITED METABOLIC DISORDERS'
VENUE: VIRTUAL HUB VIA ZOOM
DATE: 25TH, 26TH AND 27TH NOVEMBER

FESTIVAL THEME: ‘RARE THINKING’: CHANGING THE CONVERSATION FOR PEOPLE WITH INHERITED METABOLIC DISORDERS.

AUDIENCE: PEOPLE LIVING WITH INHERITED METABOLIC DISORDERS, THEIR COMMUNITIES AND ALL IMD STAKEHOLDERS

Metabolic Support UK’s ‘Rare Thinking’ Festival is a unique opportunity to gain in-depth insight into the lives of those living with Inherited Metabolic Disorders.

Our three days of themed explorations are designed to change the conversation and focus on: identity, the emergence of genomics, practical support and emerging therapies. Key disabled influencers with long term health conditions will engage with our families, elevating our advocacy offer.

Our work is set to tip the balance of what it means to live with a rare disorder and force decision makers to think in new ways.
Programme of Events

Day One  **OUR COMMON FIGHT: POWER AND IDENTITY**

An exploration of power and identity perception for people with Inherited Metabolic Disorders and how this intersects all areas of our lives. Sessions include:

**My body, my identity:** Activists living with long term health conditions discussing the intersections of healthcare, identity, diversity and human rights.

**It’s in our DNA:** As individual access to genetic makeup increases, what impact does this have on access to healthcare?

**Our voice, heard:** This session will see leading experts in advocacy and engagement sharing their advice on how to harness your lived experience to make change.

**Let’s connect:** We invite our IMD & wider community to share stories in 5 minutes or less.

**A frank conversation:** Exploring the relationship between Metabolic Support UK and pharmaceutical companies.

Speakers include:

**Angela Matthews,** *Head of Policy at Business Disability Forum* and living with Phenylketonuria (PKU)

**Sally Hatton,** *Patient advocate living with XLH and trustee of XLH UK*

**Pippa Stacey,** *Chronically ill writer and blogger*

**Laurence Woollard,** *Director of On the Pulse consultancy and patient advocate for haemophilia and rare diseases*

**Jeff Harvey,** *Disability activist living with an IMD*

**Amanda Pichini,** *Genetic Counsellor, Genomics England*

**Dr Robin Lachmann PhD FRCP,** *Consultant in Inherited Metabolic Disease*

**Lesley Greene,** *Metabolic Support UK Founder*
DAY TWO OUR EVERYDAY

Practical advice surrounding daily needs as well as an opportunity to share experiences and contribute to wider learning. Sessions include:

Your Working Life: An exploration into the barriers to employment and flexible working for those living with Inherited Metabolic Disorders and advice on how to overcome these barriers and employee rights from Advisory, Conciliation and Arbitration Service (ACAS).

A Rare Education: This session will explore the experiences and challenges of navigating the education system whilst living with an Inherited Metabolic Disorder. Our panellists will share their experiences and top tips to overcoming these challenges.

Your Money, Your Rights: What is the financial impact of living with an Inherited Metabolic Disorder and what advice and support is available? An exploration into the world of financial advice and benefits and disability rights.

Thinking About My Future: An exploration into genetic counselling and family planning.

Speakers include:

Jake Lockyer, Patient Advocate living with Hypophosphatasia (HPP)
Sarah Williams, Patient Advocate living with Adult Polyglucosan Body Disease (APBD)
Stuart Plage, Senior Advisor, ACAS
Rhiannon Dempsey, Patient Advocate living with Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
Laura Linford, Parent Advocate for a child living with Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)
Ali Hardy, Parent Advocate for a child living with HHH Syndrome
Shagufta Khan, Genetic Counsellor at Birmingham Women's and Children's NHS Foundation Trust
An opportunity for selected disorder specific communities to receive updates on the latest treatments, research and technologies. Connect with others and help us build and shape the rare disorder landscape. Sessions include:

**Connecting the CSID Community**: An opportunity for people living with Congenital sucrase-isomaltase deficiency to connect, share experiences and learn more about current research within the CSID landscape.

**Updates from the Rare Bone Alliance**: An opportunity for all those living with Rare bone disorders to connect and receive updates from Alliance members, including a 'first look' at the Rare Bone Alliance 2022 Manifesto.

**Emerging Research, Treatments and Therapies for the MMA and PA Community**: An opportunity for people living with Methylmalonic acidaemia and Propionic acidaemia to connect, share experiences and discover more about current research and emerging treatments within the MMA and PA landscape.

**Emerging Research, Treatments and Therapies for the Urea Cycle Disorder Community**: An opportunity for those living with a Urea Cycle Disorder to receive the latest news and developments on innovative treatments and research.

**Latest news and Updates from Cystinosis Foundation UK**: An opportunity for those living with Cystinosis to receive the latest news and updates from Cystinosis Foundation UK.

**Speakers include:**

**Dr Maura Corsetti**, *Clinical Associate Professor in Gastroenterology*

**Andy Wragg**, *NIHR Patient and Public Involvement Facilitator*

*Plus representatives from:*

**Brittle Bones Society**

**Hemoshear Therapeutics**

**XLH UK**

**Glasshouse Health**

**Soft Bones UK**

**Ultragenyx**

**Fibrous Dysplasia Society**

**Arcturus Therapeutics**
**Rare Thinking Festival Agenda**  
**Thursday 25th – Saturday 27th November 2021**

**Day One: Thursday 25th November**

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| 10.30-11.15am | Panel Discussion: My body, my identity | Angela Matthews, Head of Policy at Business Disability Forum and living with Phenylketonuria (PKU)  
Sally Hatton, Patient Advocate living with XLH and trustee of XLH UK  
Pippa Stacey, Chronically ill writer and blogger  
Laurence Woollard, Director of On the Pulse consultancy and patient advocate for haemophilia and rare diseases  
Chair: Kirsty Hoyle, Metabolic Support UK | A facilitated conversation between leading activists and people living with long term health conditions, discussing the intersections of healthcare, identity, diversity and human rights.  
Panellists will share their perspectives on:  
- Living with an IMD and/or long-term health condition and the impact on identity and everyday life  
- Disability/human rights and the intersections with living with an IMD/long term health condition  
- Their experiences of challenging discrimination in all areas of their lives |
| 11.30am -12.15pm | Panel Discussion: It’s in our DNA | Amanda Pichini, Genetic Counsellor, Genomics England  
Dr Robin Lachmann PhD FRCP, Consultant in Inherited Metabolic Disease  
Lesley Greene, Metabolic Support UK Founder  
Chair: Jess Doyle, Metabolic Support UK | With increasing public access to, and involvement in, genetic testing we set out to look at what this means for IMD patients.  
Topics covered:  
- What is genetic testing and the 100,000 Genomes Project?  
- What is the value of genetic testing for people living with IMD’s?  
- How does access to private genetic testing impact the future diagnosis of IMD’s? |
<p>| 1-1.45pm | Panel Discussion: Our voice, heard | Pippa Stacey, Chronically ill writer and blogger | This session will see leading experts in advocacy and patient engagement sharing their advice on how to advocate for your |</p>
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| 2-3pm   | Lightning Talks: Let’s connect                                         | **Jeff Harvey**, Disability activist living with an IMD  
**Chair: Kirsty Hoyle, Metabolic Support UK**  
More speakers announced soon! | We invite all our IMD and wider disability community to share their story in 5 minutes or less. An opportunity to connect and learn from others in the rare disorder community.  
*More details announced soon!* |
| All Day | Pre-recorded video  
A frank conversation about the relationship between Metabolic Support UK and pharmaceutical companies. | More details announced soon!                                                                                           | This short video aims to explain the relationship between Metabolic Support UK and pharmaceutical companies, and what this means for you – people living with IMD’s. Topics covered:  
- Putting the people behind the pharma – who is behind drug development?  
- Why choose rare diseases? How is the subject area selected?  
- What are the rules around patient / pharma relationship?  
- Why do Metabolic Support UK work with them?  
- How is the ‘patient voice’ represented and how does Metabolic Support UK use the information I share?  
More details announced soon! |
<p>| 4-4.45pm| Metabolic Support UK 40th Birthday Celebration and Report Launch       | <strong>Speakers</strong> will include a range of Metabolic Support UK stakeholders from the last 40 years. | This invitation only event sees the launch of our ‘40 years of change for people living with Inherited Metabolic Disorders’ report and an insight into our future plans. |</p>
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| 10.00-11.15am   | Webinar: Your working life             | Jake Lockyer, Person living with Hypophosphatasia (HPP)  
Sarah Williams, Person living with Adult Polyglucosan Body Disease (APBD)  
Stuart Plage, Senior Advisor, ACAS  
Chair: Kirsty Hoyle, Metabolic Support UK | People living with IMD’s may face challenges in their working lives. This session brings together examples of lived experience of barriers in the workplace and information on knowing your rights as an employee.  
Topics covered:  
- Living with a progressive condition and remaining in work  
- Pushed out of the workplace? What are your employer’s duties and your employee rights? |
| 11.30pm - 12.45pm | Panel Discussion: A Rare Education: advocating for accessible learning | Rhiannon Dempsey, Person living with MCADD  
Chair: Helen Morris, Metabolic Support UK  
More speakers announced soon! | This session provides peer guidance on navigating the education system to support accessible education opportunities. Hearing directly from parents, educators, students this session will support families who have members entering the education system, at all levels. |
| 1.30-2.45pm     | Panel discussion: Your money, your rights | Laura Linford, Parent of chid with VLCADD Benefits Advice Specialist, (TBC)  
Chair: Helen Morris, Metabolic Support UK  
More speakers announced soon! | Perspectives on the issues faced by people with IMD in navigating and accessing financial support plus specific advice on key issues.  
Topics covered:  
- Benefits available and how to apply for them  
- Support in completing application forms.  
- What evidence is required – how is this different for IMDs, what are the challenges?  
- Appealing decisions and where to access support.  
- Accessing equipment or supplies not covered by benefits |
### 3-4pm

**Presentation:**
Family Planning & Genetic Counselling

**Shagufta Khan, Genetic Counsellor**

*More speakers announced soon!*

Accessing genetic counselling and advice for future pregnancies, where there is a family history of an IMD. How this is facilitated and what to expect.

- What is genetic counselling?
- How to access genetic counselling
- Assessing risk for future pregnancies and family members and the support available

### Day 3: Saturday 27th November

**Theme**
My Rare Condition
An opportunity for selected disorder specific communities to receive updates on the latest treatment and technologies, connect with others and help us build and shape the rare disorder landscape.

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| 10:00am – 11:00am | Interactive Workshop: Connecting the Congenital sucrase-isomaltase deficiency Community | Andy Wragg, NIHR Patient and Public Involvement Facilitator. Dr Maura Corsetti, Clinical Associate Professor in Gastroenterology. | This session provides the opportunity for people living with CSID to connect and share experiences. Topics covered:
  - An introduction to Patient and Public Involvement.
  - Building a CSID community
  - An overview of Dr Maura Corsetti’s research study. |
| 11:30am to 12:30pm | Listening Session: Updates from the Rare Bone Disease Alliance | Representatives from: Brittle Bone Society, XLH UK, Soft Bones UK, Fibrous Dysplasia Society. | This session provides the opportunity for people living with Rare Bone Disorders to receive updates from alliance members and take a ‘first look’ at Rare Bone Alliance 2022 Manifesto. |
| 1:00pm to 2:00pm | Interactive Meeting: Emerging research, treatments and therapies for the MMA and PA Community. | Representatives from; Moderna, Hemoshear. | This session provides the opportunity for people living with Methylmalonic acidemia and Propionic acidemia to connect, share experiences and discover more about current research and emerging treatments within the MMA and PA landscape. Topics covered:
  - An update on the Moderna MrNA therapy |
| Time          | Interactive Meeting: Emerging treatments and therapies for the Urea Cycle Disorder Community. | Representatives from: Arcturus Therapeutics and Ultragenyx | An update on the Hemoshear HERO study and clinical trials  
- A Q&A panel  
This session provides an opportunity for those living with a Urea Cycle Disorder to receive the latest news and developments on innovative treatments and research.  
Topics covered:  
- An update on the Arcturus Therapeutics Lunar OTC therapy.  
- An update on the Ultragenyx DT301 OTC Therapy  
- An interactive workshop focusing on key themes from the annual survey and festival |