SSIEM
FREIBURG 2022

Genetics meets Environment

SSIEM Annual Symposium | 30 August – 2 September 2022 | Freiburg · Germany

**Dear colleagues,
welcome to the SSIEM 2022 conference in Freiburg.
Let's meet to discuss the following topics:**

Ute Spiekerkötter
Symposium President

ENVIRONMENT MEETS METABOLISM_ How do environmental factors influence health and disease, what role does the microbiome play and shouldn't we prescribe exercise in metabolic patients?

TECHNOLOGY MEETS METABOLISM_ Single cell RNA sequencing, mechanistic modelling of human metabolism and the hot topics of big data and artificial intelligence in medicine will be addressed.

ETHICS MEETS GENETICS_ The opportunities and challenges as well as the ethical aspects of whole genome screening will be explored. A lifetime's knowledge at birth? What do we actually want to know? A plenary discussion with all speakers will end the session.

REALITY MEETS GENETICS_ In this session, we want to measure what matters to rare disease patients and will focus on academia-driven medicine developments as well as on challenges and opportunities for social entrepreneurs. Sophie Hauenherm will introduce with a dance into the topic: Behind human understanding!

METABOLISM MEETS GENETICS_ We will explore how genetics changed the face of mitochondrial disease and want to understand variant pathogenicity using multi-omic pipelines. Metabolism is also influenced by the epigenetic clock, a new approach.

EPIGENETICS MEETS GENETICS_ We will learn about clinical epigenomics in metabolic diseases. Is epigenetics the 3rd dimension of phenotypic variation and disease risk? In addition a human Mendelian disorder of the DNA demethylation machinery will be introduced to us.

With all of these exciting sessions I am looking forward to meeting all of you in Freiburg!

Kind regards
Ute Spiekerkötter

SCIENTIFIC PROGRAMME

PRESIDENT & COMMITTEES

Symposium President

Ute Spiekerkötter
University Hospital of Freiburg
Freiburg, Germany

Local Organising Committee

Ute Spiekerkötter – President
Freiburg, Germany

Luciana Hannibal
Freiburg, Germany

Sarah Grünert
Freiburg, Germany

Anke Schumann
Freiburg, Germany

International Scientific Committee

Kaustuv Bhattacharya
Sydney, Australia

Helen Michelakakis
Athens, Greece

Matthias Baumgartner
Zurich, Switzerland

Eva Morava
Rochester, USA

Jim Bonham
Sheffield, UK

Kimitoshi Nakamura
Kumamoto, Japan

John Christodoulou
Parkville, Australia

Katrin Ounap
Tartu, Estonia

Roberto Giugliani
Rio Grande do Sul, Brazil

Dulce Quelhas
Porto, Portugal

Johannes Häberle
Zurich, Switzerland

David Rosenblatt
Quebec, Canada

Johan van Hove
Aurora, United States

Gajja Salomons
Amsterdam, Netherlands

Anita Inwood
Brisbane, Australia

Manuel Schiff
Paris, France

Stefan Kölker
Heidelberg, Germany

SATURDAY | 27 AUG 2022

HOTEL STADT FREIBURG

13:00 – 19:00

FAO Guideline Meeting Part I
(by invitation only)

SUNDAY | 28 AUG 2022

HOTEL STADT FREIBURG

09:00 – 13:00

FAO Guideline Meeting Part II
(by invitation only)

15:30 – 21:30

INFORM Meeting Part I
(registration at INFORM – limited places)

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Side & Administrative Meetings

Please note: This is a programme, which is subject to change without prior notice. For updated information please visit: www.ssiem2022.org

DAILY OVERVIEW | **MONDAY** | 29 AUGUST 2022

	JERUSALEM	KYOTO 1	KYOTO 2	KYOTO 3	KYOTO 4	LONDON
7:30						
8:00						
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12:00	12:00 – 16:00 Annual Meeting of the DGMet	12:00 – 14:00 NBS Collaboration Meeting – MetabERN/ ISNS/SSIEM *(by invitation only)*				
12:30						
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14:00					14:00 – 16:30 MetabERN Meeting *(by invitation only)*	
14:30						
15:00						
15:30						
16:00						16:00 – 18:00 SSIEM Council Meeting *(council members only)*
16:30	16:30 – 19:30 MetabERN SNW Amino and Organic acids-related Disorders – AOA *(by invitation only)*		16:30 – 19:30 MetabERN SNW Disorders – PD *(by invitation only)*	16:30 – 19:30 MetabERN SNW Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders – C-FAO *(by invitation only)*	16:30 – 19:30 MetabERN SNW Lysosomal Storage Disorders – LSD *(by invitation only)*	
17:00						
17:30						
18:00		18:00 – 19:30 MetabERN SNW PM-MD *(by invitation only)*				
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Side & Administrative Meetings

MEETING ROOM 1	MEETING ROOM 4	MEETING ROOM 5	HOTEL STADT FRBG
			07:30 – 21:00 INFORM Meeting Part II *(registration at INFORM – limited places)*
11:00 – 13:45 Annual E-IMD Members Meeting *(by invitation only)*			
	16:30 – 19:30 MetabERN SNW Congenital Disorders of Glycosylation and Disorders of Intracellular Trafficking – CDG *(by invitation only)*	16:30 – 19:30 MetabERN SNW Disorders of Neuromodulators and Other Small Molecules – NOMS *(by invitation only)*	
18:00 – 22:00 ERNDIM Board of Trustees Meeting *(by invitation only)*			

DAILY OVERVIEW | **TUESDAY** | 30 AUGUST 2022

	FREIBURG	JERUSALEM	PORTO	KYOTO 1	KYOTO 2	KYOTO 3
7:30						
8:00						
8:30						
9:00	09:00 – 12:00 SSIEM Adult Group Meeting *(open to all participants)*	09:00 – 10:30 ERNDIM Workshop: DPT Netherlands *(by invitation only)*	09:00 – 12:30 Nutrition and Dietetic Session *(open to all participants)*	09:00 – 10:30 ERNDIM Workshop: DPT Czech Republic *(by invitation only)*	09:00 – 10:30 ERNDIM Workshop: DPT Switzerland *(by invitation only)*	09:00 – 10:30 ERNDIM Workshop: DPT France *(by invitation only)*
9:30						
10:00						
10:30						
11:00		11:00 – 12:30 ERNDIM Workshop *(open to all participants)*				
11:30						
12:00				12:00 – 13:30 EHOD Members Meeting *(by invitation only)*		
12:30						
13:00	12:45 – 13:45 Satellite Symposia	12:45 – 13:45 Satellite Symposia	12:45 – 13:45 Satellite Symposia		12:45 – 13:45 SSIEM Adult Group Business Meeting *(for all members involved in adult metabolic medicine)*	
13:30						
14:00	14:00 – 14:40 Opening Ceremony					
14:30						
15:00	14:40 – 15:45 Environment meets Metabolism					
15:30						
16:00						
16:30	16:15 – 18:00 Technology meets Metabolism					
17:00						
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18:00	18:00 – 18:30 Garrod Award Lecture					
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Networking | Plenary Session | Satellite Symposia | Poster | Side & Administrative Meetings

KYOTO 4

09:00 – 10:30
ERNDIM Workshop: DPT UK *(by invitation only)*

12:45 - 13:45
Speed Mentoring

LONDON

09:00 – 10:30
ERNDIM Workshop: NCSF participants' meeting
(NCSF participants only)

MEETING ROOM 1

09:00 – 12:00
GalNet Symposium
(by invitation only)

MEETING ROOM 4

10:00 – 12:00
ETAC Meeting
(by invitation only)

MEETING ROOM 5

09:00 – 11:00
EHOD Executive Board Meeting
(by invitation only)

MEETING ROOM 6

09:00 – 12:00
JIMD and JIMD Reports – Editorial Meeting
(by invitation only)

MEETING ROOM 7

09:00 – 12:30
Molybdenum cofactor deficiency Guideline Development Group meeting
(by invitation only)

POSTER EXHIBITION

14:00 – 18:30
Poster & e-Poster

FOYER

18:30 – 21:30
Welcome Reception

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DAILY OVERVIEW | **WEDNESDAY** | 31 AUGUST 2022

	FREIBURG	JERUSALEM	PORTO	KYOTO	LONDON	MEETING ROOM 1
7:30		07:30 – 08:30 Satellite Symposia	07:30 – 08:30 Satellite Symposia	07:30 – 08:30 Satellite Symposia	07:30 – 08:30 IOC Meeting	07:30 – 08:30 Speed Mentoring
8:00						
8:30						
9:00	08:45 – 10:15 New Therapies in Lysosomal Disorders	08:45 – 10:15 Organic Acidurias	08:45 – 10:15 Glycosylation and Carbohydrate Disorders	08:45 – 10:45 SSIEM Nurses Meeting *(open to all participants)*	08:45 – 10:15 Patient Representative Meeting	
9:30						
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11:00	10:45 – 12:45 Ethics meets Genetics		10:45 – 12:45 SSIEM-DG Meeting *(open to all participants)*			
11:30						
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13:00		13:00 – 13:30 SSIEM Advisory & Council Members Meeting (only SSIEM Advisory Council & Council Members)	12:45 – 13:15 SSIEM-DG Executive Committee Meeting *(by invitation only)*			
13:30		13:30 – 14:30 SSIEM Annual General Assembly (only SSIEM Members)				
14:00						
14:30						
15:00	14:45 – 16:15 Mechanisms and Markers in Lysosomal Disorders	14:45 – 16:15 New Perspectives in Phenylketouria	14:45 – 16:15 Disorders of Fat Metabolism	14:45 – 16:15 Novel Diagnostic Technologies		
15:30						
16:00						
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17:00	16:45 – 18:35 Reality meets Metabolism					
17:30						
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19:00		18:45 – 19:45 Satellite Symposia	18:45 – 20:15 Satellite Symposia			
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Networking | Plenary Session | Satellite Symposia | Side & Administrative Meetings
Parallel Session | New Formats | Poster

	SCIENTIFIC THEATRE	POSTER EXHIBITION	CITY CENTRE
7:30		07:30 – 20:15 Poster & e-Poster	
8:00			
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13:00	13:00 – 13:45 The Needs of Patients		
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19:00	18:45 – 20:15 Presentation of high ranked posters	18:45 – 20:15 Poster Walk	
19:30			
20:00			
20:30			20:30 – 00:00 Young SSIEM Evening
21:00			
21:30			
22:00			
22:30			

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DAILY OVERVIEW | **THURSDAY** | 1 SEPTEMBER 2022

	FREIBURG	JERSUALEM	PORTO	KYOTO	MEETING ROOM 1	GALLERY ROOM 1
7:30		07:30 – 08:30 Satellite Symposia	07:30 – 08:30 Satellite Symposia	07:30 – 08:30 Satellite Symposia	07:30 – 08:30 Speed Mentoring	
8:00						
8:30						
9:00	08:45 – 10:30 Metabolism meets Genetics					
9:30						
10:00						
10:30						10:30 – 11:00 Coffee with the JIMD Editors
11:00	11:00 – 12:30 Gene Therapy Clincial Trials	11:00 – 12:30 Urea Cycle Disorders	11:00 – 12:30 Mitochondrial Disorders	11:00 – 12:30 Disorders of Vitamins and Cofactors		
11:30						
12:00						
12:30					12:30 – 14:30 JIMD and JIMD Reports – Editorial Meeting *(by invitation only)*	12:30 – 14:00 Alumni Café
13:00	12:45 – 13:45 Satellite Symposia	12:45 – 13:45 Satellite Symposia	12:45 – 13:45 Satellite Symposia	12:45 – 13:45 Satellite Symposia		
13:30						
14:00	14:00 – 15:30 Gene and innovative Therapies	14:00 – 15:30 Amino Acid Disorders	14:00 – 15:30 Neurometabolic Disorders	14:00 – 15:30 Novel Disease Insights		
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Networking | Plenary Session | Satellite Symposia | Side & Administrative Meetings
Parallel Session | New Formats | Poster

THURSDAY | 1 SEPT. 2022

SCIENTIFIC THEATRE

12:45 – 13:00
Science Slam

FOYER

15:30 – 18:30
Networking Activities

POSTER EXHIBITION

07:30-15:30
Poster & e-Poster

KONZERTHAUS

19:30 – 00:00
Networking Evening with Dinner Speech

FRIDAY | 2 SEPTEMBER 2022

FREIBURG

08:45 – 10:30
Epigenetics meets Genetics

11:00 – 11:45
Komrower Lecture

11:45 – 12:45
Special Awards Session

12:45 – 13:15
Awards

MEETING ROOM 1

07:30 – 08:30
Speed Mentoring

POSTER EXHIBITION

07:30-13:15
Poster & e-Poster

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15:30 – 21:30 | INFORM Meeting Part I *(registration at INFORM – limited places)*
Hotel Stadt Freiburg | Side & Administrative Meetings

International Network for Fatty Acid Oxidation Research and Management
Moderation: Vockley, Jerry (Pittsburgh, USA)

15:30 **Registration**
16:30 **Welcome** | *Vockley, Jerry (Pittsburgh, USA)*
16:45 **»Philosophical«: What have we learned, what can we expect for the future**
Vianey-Saban, Christine (Lyon, France)
17:45 **Research Discussion**

18:15 **| Break**

19:00 **Networking Reception & Poster Presentation**

07:30 – 21:00 | INFORM Meeting Part II *(registration at INFORM – limited places)*
Hotel Stadt Freiburg | Side & Administrative Meetings

International Network for Fatty Acid Oxidation Research and Management
Moderation: Vockley, Jerry (Pittsburgh, USA)

07:30 **Registration**
08:30 **Confirmatory Genetic Analysis, when is a positive screen a diagnosis**
Vockley, Jerry (Pittsburgh, USA)
09:00 **Initiation of Treatment / Pre-symptomatic Treatment**
Burton, Barbara (Chicago, USA)
09:30 **Junior Investigator Presentation**
09:45 **Junior Investigator Presentation**

10:00 **| Break**

10:30 **Key nutrition principles for management of FAOD** | *Singh, Rani (Atlanta, USA)*
11:00 **Data on energy balance and calculating total energy needs in subjects with FAODs using the C7 data** | *Gillingham, Melanie (Portland, USA)*
11:30 **Junior Investigator Presentation**
11:45 **Junior Investigator Presentation**
12:00 **Panel Discussion**

12:30	**| Lunch / Announce Poster Winners; MitoAction & INFORM Family Luncheon; Patient and Family Discussion |** *Spiekerkötter, Ute (Freiburg, Germany) & Vockley, Jerry (Pittsburgh, USA)*
14:00	**Update on Cardiomyopathy in FOADs** | *Chatfield, Kathryn C. (Aurora, USA)*
14:30	**Cardiolipin Metabolism and the Heart** | *Ruohola-Baker, Hannele (Seattle, USA)*
15:00	**Junior Investigator Presentation**
15:15	**Junior Investigator Presentation**
15:30	**| Break**
15:45	**Life with a child affected by a fatty acid oxidation defect: Impact on different areas of life, psychological aspects and coping strategies** *Grünert, Sarah (Freiburg, Germany) & Thiel, Maren (Berlin, Germany)*
16:15	**Natural History of Retinopathy / LCHAD and the Eye** | *Pennesi, Mark (Portland, USA)*
16:45	**Case study of Physical Therapy for LCHAD deficiency** | *Tucker, Pamela (Syracuse, USA)*
17:00	**Junior Investigator Presentation**
17:15	**Panel Discussion / Summary & Closing remarks**
17:45	**| Break**
18:00	**INFORM Committee Meeting**
19:00	**| Break**
19:30	**INFORM Night out in Freiburg**

Hans-Albert Stechl

Lukewarm Beef Asparagus Salad

This salad is a main course. At least when you do not just eat a piece of baguette with it, but a nice portion of roast potatoes.

If you cook the brisket with the bones, two bay leaves, a carrot, a piece of leek, half a bunch of parsley, a piece of celeriac, an onion, a clove of garlic and a heaped teaspoon of peppercorn, the most welcome by-product is 2 litres of the finest meat broth. The cooking time for the meat is about 2 hours. The water should not boil wildly, but simmer gently. Use a pointed fork to determine whether the meat is cooked. If the fork penetrates the meat easily, it is done. The old question of whether to put the meat in cold or hot water can be quickly settled: If you want the meat to be as rich as possible, put it in cold water. In

this case, the meat will be leached out a little better when the water is heated slowly. If the focus is on a piece of meat that is as juicy as possible, put it in boiling water.

We combine both cooking methods: Put the bones and all the other ingredients into cold water. Place the meat into the pot as soon as the water boils. Once cooked, cut the meat into small thin slices, put it in a pot and cover it with broth. This prevents it from drying out. Heat it again in the broth shortly before serving.

Asparagus. We recommend asparagus tips or broken asparagus, because they will be cut into pieces anyway. Cook the asparagus until tender to the bite. Clean the baby carrots and likewise cook them until al dente. If a little of the green remains on the carrots, it does not affect the taste, and it looks nice. Cook the sugar snap peas in boiling water for 2 minutes and then rinse with cold running water. Peel the red onion and cut into very fine strips. Clean the radishes and slice them not too thinly. For the herb vinaigrette, mix the mustard, vinegar, meat stock, salt, pepper and sugar until the mustard has dissolved, then add the oil. Chop the parsley, chives and lovage finely and stir into the vinaigrette. Heat the meat in the stock, strain through a sieve, mix loosely with all the ingredients and the vinaigrette and arrange on a platter.

Ingredients *(for 4 servings)*

1 kg lightly marbled brisket of beef
1 handful of beef bones
2 bay leaves
1 carrot
½ leek
1 small piece of celeriac
½ bunch parsley
1 small onion
1 clove of garlic
1 heaped teaspoon peppercorns

500 g asparagus tips or broken asparagus
½ bunch carrots (baby carrots)
100 g sugar snap peas
1 small red onion
½ bunch radishes

Vinaigrette
1 heaped teaspoon hot mustard
3 tablespoons vinegar
3 teaspoons meat stock
salt and pepper
1 pinch of sugar
5 tablespoons olive oil
½ bunch parsley
½ bunch chives
½ bunch lovage

For more recipes please visit: www.freiburger-marktkalender.de

09:00 – 11:00 | SSIEM Adult Group Meeting *(open to all participants)*
Freiburg | Side & Administrative Meetings

Update on porphyria
Moderation: Mochel, Fanny (Paris, France)

09:00 **Introduction and welcome**

09:05 **Interference RNA treatment for acute porphyria**
Sardh, Eliane (Stockholm, Sweden)

09:35 **Treatment of erythropoietic protoporphyria** | *Langendonk, Janneke (Rotterdam, Netherlands)*

10:05 **Liver complications in porphyrias** | *Ventura, Paolo (Modena, Italy)*

10:35 **Concluding remarks**

10:45 **| Break**

11:00 **Epidemiological Trends and Outcomes of Children, Adolescents, and Adults Hospitalized with Inherited Metabolic disorders : A Population Based Cohort Study** | *Hauser, Stephanie (Aarau, Switzerland)*

11:15 **Plasma globotriaosylsphingosine correlates strongly with disease severity in untreated Fabry disease patients: A tool to aid clinical decision making** | *van der Veen, Sanne (Amsterdam, Netherlands)*

11:30 **Effects of high versus low Phe intake on neurocognitive functioning and wellbeing in adults with PKU; The Phe eat or diet study** | *Wagenmakers, Margreet (Rotterdam, Netherlands)*

11:45 **Alpha-mannosidosis diagnosed in a 47-year-old male: the importance of re-visiting undiagnosed patients** | *Nurse, James (Southampton, UK)*

09:00 – 12:30 | Nutrition and Dietetic Session *(open to all participants)*
Porto | Side & Administrative Meetings

Moderation: Dianin, Alice (Verona, Italy), Lang, Frauke (Mainz, Germany)

09:00 **The gut microbiome in inherited metabolic disorders: the influence of the diet** | *Timmer, Corrie (Amsterdam, Netherlands)*

09:30 **The changing face of PKU - the challenges of nutrition and dietetics in the era of new treatment approaches** | *Rohde, Carmen (Leipzig, Germany)*

10:00 **Overcoming nutritional problems in IEM post-transplant** | *Pedro, Temitope (Stanford, USA)*

10:30 | **Break**

10:45 **Efficacy and safety of empagliflozin in glycogen storage disease type Ib** | *Grünert, Sarah (Freiburg, Germany)*

11:15 **Pregnancies in phenylketonuria** | *Lier, Dinah (Reutlingen, Germany)*

11:30 **Pregnancies in other inborn errors of metabolism** | *Green, Diane (Salford, UK)*

11:00 – 12:30 | ERNDIM Workshop *(open to all participants)*
Jerusalem | Side & Administrative Meetings

Moderation: Artuch, Rafael (Esplugues de Llobregat, Spain)

11:00 **Chair's Update** | *Artuch, Rafael (Esplugues de Llobregat, Spain)*

11:10 **Common DPT sample** | *Croft, Joanne (Sheffield, UK)*

11:20 **Biomarkers for neurotransmitter diseases** | *Heales, Simon (London, UK)*

11:50 **Biomarkers for mitochondrial diseases** | *Horvath, Rita (Cambridge, UK)*

12:20 **Open discussion: Future directions** | *Artuch, Rafael (Esplugues de Llobregat, Spain)*

14:00 – 14:40 | Opening Ceremony
Freiburg | Opening

14:00 **Eroica** | *Children*

14:08 **Welcome Speech** | *Spiekerkötter, Ute (Freiburg, Germany)*

14:23 **Everyone** | *Matrix*

14:30 **The power of humanity** | *Matrix and Children*

14:40 – 15:45 | Environment meets Metabolism
Freiburg | Plenary Session 1

Chairs: Bhattacharya, Kaustuv (Sydney, Australia), Kölker, Stefan (Heidelberg, Germany)

14:40 **Introduction into the session** | *Grünert, Sarah (Freiburg, Germany)*

14:45 **Genetics and the microbiome** | *Ley, Ruth (Tübingen, Germany)*

15:15 **Exercise testing and prescription in metabolic diseases** | *Broderick, Carolyn (Sydney, Australia)*

15:45 – 16:15 | **Coffee Break**

16:15 – 18:00 | Technology meets Metabolism
Freiburg | Plenary Session 2

Chairs: Michelakakis, Helen (Athens, Greece), Van Hove, Johan (Aurora, USA)

16:15 **Introduction into the session** | *Schuman, Anke (Freiburg, Germany)*

16:25 **Single cell RNA sequencing: understanding liver cell differentiation and plasticity in health and disease** | *Heikenwälder, Mathias (Heidelberg, Germany)*

16:55 **Mechanistic modeling of human metabolism: monogenic and polygenic disorders** | *Fleming, Ronan M.T. (Galway, Ireland)*

17:25 **Big data and artificial intelligence in medicine: the good, the bad and the ugly** | *Lovis, Christian (Genf, Switzerland)*

18:00 – 18:30 | Garrod Award Lecture
Freiburg | Award Lecture

Chair: Rahman, Shamima (London, UK)

18:00 **Delineating the clinical spectrum of isolated methylmalonic acidurias: cblA and mut** | *Hörster, Friederike (Heidelberg, Germany)*

18:30 – 21:00 | Welcome Reception
Foyer | Networking

Hans-Albert Stechl

Potato, leek and ham quiche

Saying that the served food is made up of leftovers rarely causes a storm of enthusiasm at the table. After all, it still sounds like leftovers.

That's why I prefer to talk about the culinary harmony of food items that are only available in such small quantities that if used individually, do not serve four people. Our fridge had two and a half slices of cooked ham from breakfast of the day before, two leeks, two potatoes, three eggs, half a cup of cream and a well-hardened piece of Parmesan cheese.

Boil the potatoes in their skins until soft, peel and cut into thin slices. Cut off the green parts of the leek that are no longer dewy, as well as the root end. Cut the remaining light-coloured stalk into not too thick

slices. Rinse well in a sieve to remove any residual garden soil. Then steam the slices in a pan with butter over moderate heat for 10 minutes.

Cut the ham into thin slices. Whisk the eggs with the cream, grate the Parmesan, mix with the whisked eggs, salt, pepper and season with nutmeg. Mix the leek with the ham mixture.

Line a 26-centimetre-diameter pan with shortcrust pastry and form a rim about 3 centimetres high. Poke the base several times with a fork.

Cover the base evenly with the potato slices. Sprinkle marjoram - fresh or dried - over the potato slices. Spread the leek and ham mixture evenly over the potato slices. Finally, pour the egg and cream mixture over the top so that everything is nicely moistened. Put the quiche in the oven preheated to 200 degrees (top and bottom heat, middle shelf) and bake for about 40 minutes.

For the shortcrust pastry: If you do not have time, use a ready-made one from a roll. Look carefully - there are salted and sugared ones. However, kneading the shortcrust pastry yourself is anything but rocket science. Knead the flour with your hands (spelt flour works very well here) with room temperature butter cut into pieces, a good pinch of salt and water until you have a smooth dough. Form the dough into a ball, wrap it in kitchen foil and put it in the fridge for 30 minutes. Then roll out a thin dough.

Ingredients *(for 4 servings)*

2–3 slices cooked ham
2 leeks
2 medium potatoes (firm or floury)
3 eggs
½ cup cream
50 g grated parmesan (another leftover hard cheese works as well)
Salt and pepper
Nutmeg
Marjoram
Butter to steam the leeks

Shortcrust pastry

1 roll of pre-made salted pastry or:
200 g spelt flour
100 g room temperature butter
2 tablespoons cold water
1 good pinch of salt
Kitchen foil

For more recipes please visit: www.freiburger-marktkalender.de

08:45 – 10:15 | New Therapies in Lysosomal Disorders
Freiburg | Parallel Session 1A

Chairs: Giugliani, Roberto (Porto Alegre, Brazil), Muschol, Nicole (Hamburg, Germany)

08:45 **Retargeting phenylbutyrate, ursodeoxycholic acid, pyrimethamine and betaine for beta-glucocerebrosidase recovery in gaucher disease fibroblasts resulting from homozygous p.L483P mutation** | *Kiliç, Ayse (Ankara, Turkey)*

09:00 **Four-year real-world effectiveness of eliglustat in treatment-naive and switch patients enrolled in the International Collaborative Gaucher Group (ICGG) Gaucher Registry** | *Mistry, Pramod (New Haven, USA)*

09:15 **AZ-3102, a novel brain-penetrant small molecule, significantly improves survival in Sandhoff disease mice** | *Walia, Jagdeep (Kingston, Canada)*

09:30 **The first successful in utero enzyme replacement therapy treatment of a child with CRIM negative infantile-onset Pompe disease** | *Cohen, Jennifer L. (Durham, USA)*

09:45 **Interim 73-week cohorts A, B, and C results of a Ph1/2 study of intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II** *Muenzer, Joseph (Chapel Hill, USA)*

10:00 **RGX-121 gene therapy for the treatment of severe mucopolysaccharidosis type II (MPS II): Interim analysis of data from a Phase 1/2 study** *Giugliani, Roberto (Porto Alegre, Brazil)*

08:45 – 10:15 | Organic Acidurias
Jerusalem | Parallel Session 1B

Chairs: Mühlhausen, Chris (Göttingen, Germany), Scholl-Bürgi, Sabine (Innsbruck, Austria)

08:45 **Altered glutamine anaplerosis in MMUT deficiency** | *Traversi, Florian (Zurich, Switzerland)*

09:00 **Different urinary biomarker patterns in a large cohort of patients with classical organic acidurias of branched-chain amino acid metabolism** *Köpfer, Felix (Heidelberg, Germany)*

09:15 **The liver in methylmalonic and propionic acidemias: are there risk factors for cancer?** | *Brassier, Anaïs (Paris, France)*

09:30 **A network medicine approach identifies key TCA cycle enzymes as potential therapeutic targets in organic acidemias** | *Piper, Zina (Hamburg, Germany)*

09:45 **Cognitive functions are not necessarily spared in early diagnosed individuals with glutaric aciduria type 1 – a national prospective study over 20 years** *Märtner, Eva Marie Charlotte (Heidelberg, Germany)*

10:00 **Newborn screening and disease variants predict neurological outcome in isovaleric aciduria** | *Mütze, Ulrike (Heidelberg, Germany)*

08:45 – 10:15 | Glycosylation and Carbohydrate Disorders
Porto | Parallel Session 1C

Chairs: Morava, Eva (Rochester, USA), Santer, Rene (Hamburg, Germany)

08:45 **Sodium D,L-3-hydroxybutyrate in the treatment of GLUT1 deficiency syndrome** | *Bernhardt, Isaac (Auckland, New Zealand)*

09:00 **Anaplerotic therapy using Triheptanoin for patients with glycogen storage disease type I (GSD I): clinical trial results** | *El-Gharbawy, Areeg (Durham, USA)*

09:15 **Investigating metabolic adaptations in PMM2-CDG** | *Radenkovic, Silvia (Rochester, USA)*

09:30 **Functional platforms for the characterization of PMM2-CDG clinical variants** | *Falquina, Cristina Segovia (Madrid, Spain)*

09:45 **Oral sialic-acid supplementation in NANS-CDG: Results of a single center, open label, observational pilot study** | *van Karnebeek, Clara (Amsterdam, Netherlands)*

10:00 **Cellular fluxomics to study sugar metabolism in Galactosemia and Congenital Disorders of Glycosylation** | *Noga, Marek (Nijmegen, Netherlands)*

08:45 – 10:15 | Patient Representative Meeting
London | New Formats

08:45 – 10:45 | SSIEM Nurses Meeting *(open to all participants)*
Kyoto | Side & Administrative Meetings

Moderation: Inwood, Anita (South Brisbane, Australia)

08:45 **Introduction and welcome** | *Meyer, Marie-Louise (Freiburg, Germany)*

09:00 **Comparison of inpatient length of stay (LOS) at a quaternary paediatric hospital** | *Dalkeith, Troy (Sydney, Australia)*

09:20 **Clinical nurse specialist role and transition: The model of care for transition in Manchester and the role of the clinical nurse specialist** | *Hutton, Rebekah (Manchester, UK)*

09:40 **A single metabolic centres experience of continuous glucose monitoring (CGM) for monitoring of hypoglycaemia** | *Smith, Katie (Bristol, UK)*

10:00 **NP/CNC/RN role presentation: The value of clinical nurse specialists and nurse practitioners in a metabolic service** | *Annemarie De Vreugd (Nijmegen, Netherlands)*

10:20 **An 18-month-old female with ornithine transcarbamylase (OTC) deficiency who presented with hyperammonaemia which progressed to acute liver failure** | *Inwood, Anita (South Brisbane, Australia)*

10:40 **Summary and close** | *Meyer, Marie-Louise (Freiburg, Germany)*

10:15 – 10:45 | **Break**

10:45 – 12:45 | Ethics meets Genetics
Freiburg | Plenary Session 3

Chairs: Bonham, James Robert (Sheffield, UK), Spiekerkötter, Ute (Freiburg, Germany)

10:45 **Introduction into the session** | *Spiekerkötter, Ute (Freiburg, Germany)*

10:55 **Newborn screening by whole genome sequencing: opportunities and challenges** | *Bick, David (London, UK)*

11:20 **Ethics of big data and whole genome screening: how to make chicken from chicken salad?** | *Krones, Tanja (Zurich, Switzerland)*

11:45 **A lifetime's knowledge at birth? What do we actually want to know?** *Hopkins, Henrietta (London, UK)*

12:10 **Plenary discussion moderated by J. Bonham, U. Spiekerkötter** | *Bick, David (London, UK), Gross, Edith (Paris, France), Hopkins, Henrietta (London, UK), Krones, Tanja (Zurich, Switzerland)*

10:45 – 12:45 | SSIEM-DG Meeting *(open to all participants)*
Porto | Side & Administrative Meetings

Moderation: Rocha, Júlio César (Lisboa, Portugal)

10:45 **Welcome and business meeting**

11:15 **Dietary management of homocystinuria (HCU caused by cystationine beta-synthase deficiency: Perspectives from a global cohort of metabolic dietitians** | *Starin, Danielle (Washington, USA)*

11:30 **A three-year longitudinal study comparing bone mass, density and geometry measured by DXA, pQCT and bone turnover markers in children with PKU taking either L amino acid or glycomacropeptide protein substitutes** | *Daly, Anne (Birmingham, UK)*

11:45 **Normalization of intact protein intake among adults with phenylketonuria after sustained Pegvaliase treatment** | *Singh, Rani (Atlanta, USA)*

12:00 **Anita MacDonald lecture: What can we expect from nutrition in the next decade?** | *Singh, Rani (Atlanta, USA)*

12:30 **Conclusion**

12:45 – 14:45 | Lunch & Poster Viewing

13:00 – 13:45 | The Needs of Patients
Scientific Theatre | New Formats

13:00 – 13:30 | SSIEM Advisory & Council Members Meeting
Jerusalem | Side & Administrative Meetings (only SSIEM Advisory Council & Council Members)

13:30 – 14:30 | SSIEM Annual General Meeting
Jerusalem | Side & Administrative Meetings (only SSIEM Members)

14:45 – 16:15 | Mechanisms and Markers in Lysosomal Disorders
Freiburg | Parallel Session 2A

Chairs: Eyskens, Francois (Antwerp, Belgium), Van der Ploeg, Ans (Rotterdam, Netherlands)

14:45 **Lyso-Gb3 elicits a proteotoxic effect on a neuronal cell model**
Heywood, Wendy (London, UK)

15:00 **Next-generation deep plasma proteomics reveals systemic and tissue-specific remodeling in Fabry Disease** | *Bekri, Soumeya (Rouen, France)*

15:15 **Quantitative nuclear magnetic resonance spectroscopy-based metabolomics of urine samples in metachromatic leukodystrophy: identifying indicators for neurodegeneration and disease progression** | *Laugwitz, Lucia (Tübingen, Germany)*

15:30 **LC-MSMS sulfatides measurement in dried blood spots for the diagnosis of metachromatic leukodystrophy** | *Pettazzoni, Magali (Bron, France)*

15:45 **Neurofilament light as a biomarker for involvement of the brain in classic infantile Pompe patients** | *Mackenbach, Maarten (Rotterdam, Netherlands)*

16:00 **Bi-allelic variants in VPS16, encoding a subunit of HOPS/CORVET complexes, cause a mucopolysaccharidosis-like disease** | *Cayuela, Jorge Asin (Gothenburg, Sweden)*

14:45 – 16:15 | New Perspectives in Phenylketonuria
Jerusalem | Parallel Session 2B

Chairs: Hennermann, Julia B. (Mainz, Germany), Karall, Daniela (Innsbruck, Austria)

14:45 **Characterization of a humanized PKU mouse carrying the frequent splicing variant c.1066-11G>A** | *Desviat, Lourdes (Madrid, Spain)*

15:00 **Cognitive deficits emerge in early-treated adult PAH-deficient mice following discontinuation of pegvaliase therapy** | *Harding, Cary O. (Portland, OR, USA)*

15:15 **Investigation of the phenylalanine hydroxylase proteostasis network reveals potential therapeutic targets for phenylketonuria** | *Haupt, Luka Janina (Hamburg, Germany)*

15:30 **Searching for new potential biomarkers in adults with Phenylketonuria**
van Wegberg, Annemiek (Groningen, Netherlands)

15:45 **A long non-coding RNA (lncRNA) transcript HULC regulates phenylalanine hydroxylase activity and could act as a new therapeutical agent in phenylketonuria** | *Feillet, François (Nancy, France)*

16:00 **A CRISPR/Cas9 genome-edited PAH-deficient cell line for studying PKU**
Koppes, Erik (Pittsburgh, USA)

14:45 – 16:15 | Disorders of Fat Metabolism
Porto | Parallel Session 2C

Chairs: Vockley, Jerry (Pittsburgh, USA), Wanders, Ronald (Amsterdam, Netherlands)

14:45 **Biochemical studies in fibroblasts to interpret variants of unknown significance in the ABCD1 gene** | *Ferdinandusse, Sacha (Amsterdam, Netherlands)*

15:00 **Upregulation of glycogen cycling with a triheptanoin diet replenishes glycogen stores in very long chain acyl-CoA dehydrogenase deficient mice (VLCAD-/-)** | *Nurjanah, Siti (Freiburg, Germany)*

15:15 **Thermo-sensitive mitochondrial trifunctional protein deficiency presenting with episodic myopathy** | *Schwantje, Marit (Utrecht, Netherlands)*

15:30 **Restoring succinyllysine antigenic signal and improving O2 consumption of CPT II deficient cells treated with anaplerotic compounds** | *Seminotti, Bianca (Pittsburgh, USA)*

15:45 **Plasma metabolomics during anabolic conditions among subjects with a fatty acid oxidation disorder compared to normal controls** | *Gillingham, Melanie (Portland, USA)*

16:00 **Systemic corticosteroids for the treatment of acute episodes of rhabdomyolysis in lipin-1-deficient patients** | *de Lonlay, Pascale (Paris, France)*

14:45 – 16:15 | Novel Diagnostic Technologies
Kyoto | Parallel Session 2D

Chairs: Ribes, Antonia (Barcelona, Spain), Vaz, Frederic (Amsterdam, Netherlands)

14:45 **Analysis of urinary oligosaccharide excretion patterns by UHPLC/HRAM-MS for rapid detection of oligosaccharidoses** | *Hagemeijer, Marne (Rotterdam, Netherlands)*

15:00 **Discovery of new plasma biomarkers for Sjögren Larsson syndrome by untargeted lipidomics** | *Vaz, Frederic (Amsterdam, Netherlands)*

15:15 **Unmasking the functional impact of variants of uncertain significance using knock-in cell lines generated by CRISPR/Cas9** | *Muñoz-Pujol, Gerard (Barcelona, Spain)*

15:30 **Liver-on-a-tube: Hollow fiber membrane technology to study liver metabolism and disease** | *Lehmann, Vivian (Utrecht, Netherlands)*

15:45 **Multiplexing complexome profiling to foster routine protein complex profiling in medical research** | *Guerrero-Castillo, Sergio (Hamburg, Germany)*

16:00 **Expanded phenotyping by microscopic imaging** | *Roels, Frank (Gent, Belgium)*

16:15 – 16:45 | **Coffee Break**

16:45 – 18:35 | Reality meets Metabolism
Freiburg | Plenary Session 4

Chairs: Häberle, Johannes (Zurich, Switzerland), Inwood, Anita (Brisbane, Australia)

16:45 **Introduction into the session** | *Grünert, Sarah (Freiburg, Germany)*

16:55 **Sophie Hauenherm – Behind human understanding** | *Hauenherm, Sophie (Dresden, Germany)*

17:05 **Measuring what matters to rare disease patients – patient-reported outcomes** | *Huemer, Martina (Bregenz, Austria)*

17:35 **Academia-driven medicine development: towards new public-private partnerships** | *Hollak, Carla (Amsterdam, Netherlands)*

18:05 **Rare diseases: challenges and opportunities for social entrepreneurs** *Sireau, Nicolas (Cambridge, UK)*

18:45 – 20:15 | Poster Walk | Poster Exhibition

18:45 – 20:15 | High ranked Posters
Scientific Theatre | New Formats

20:30 – 00:00 | Young SSIEM Evening
Markthalle | Networking

Hans-Albert Stechl

Grandma's apple pie

This apple pie warms the soul and it keeps us meaningfully busy in the kitchen for 2 hours.

It all starts with a shortcrust pastry: add flour, sugar, salt, water and refrigerator-cold butter cut into small pieces into a bowl. Many swear by margarine instead of butter, not for reasons of economy, but because margarine supposedly makes the dough a touch crumblier. Since tasty premium margarine is available nowadays, this variant is great to use. Knead the ingredients with your hands until everything is smoothly combined. Form the dough into a ball, cover and chill in the refrigerator for 30 minutes. Peel the apples, cut into eighths, and remove the core, place in a bowl and sprinkle generously with lemon

juice. Lay out a piece of baking paper, place the dough ball in the centre, dust with flour and roll out with a rolling pin until you have a round plate with a diameter slightly larger than the diameter of the baking pan. For a 26-centimetre baking pan, the dough layer should measure about 35 centimetres.
Transfer the pastry sheet together with the baking paper into the pie dish and press down on the bottom and sides. Cut away any excess dough on the edges. Cover the base with the apple slices. Brush the apples with melted butter, sprinkle lightly with sugar and cinnamon. Bake for 25 minutes in an oven preheated to 180 degrees Celsius (place the pie crust below the oven's middle shelf, so that the base bakes well).

Icing: Beat the sugar, vanilla sugar and eggs in a bowl with a mixer (whisk inserts). This takes at least five minutes, until the mixture is foamy and takes on a light colour. Melt the butter in a saucepan and add the cream, then heat, but do not bring to a boil. Pour the cream into the foamy sugar and egg mixture while stirring, and then pour the glaze over the apples.

Bake the pie for a further 20 minutes with the last 5 minutes set with bottom heat only.

Take the pie out, sprinkle lightly with sugar and caramelise by returning the pie to the oven with heat set on high.

Ingredients

Shortcrust pastry
250 g flour
125 g butter or margarine
50 g sugar
1 pinch of salt
2 tablespoons cold water

Topping
1 kg crisp apples
Some lemon juice
50 g butter
Cinnamon powder
Sugar

Icing
60 g sugar
1 tablespoon vanilla sugar
2 eggs
30 g butter
200 g cream
Sugar for caramelising

For more recipes please visit: www.freiburger-marktkalender.de

08:45 – 10:30 | Metabolism meets Genetics
Freiburg | Plenary Session 5

Chairs: Freisinger, Peter (Reutlingen, Germany), Ounap, Katrin (Tartu, Estonia)

08:45 **Introduction into the session** | *Schumann, Anke (Freiburg, Germany)*

08:55 **How genetics changed the face of mitochondrial disease**
Tyynismaa, Henna (Helsinki, Finland)

09:25 **Understanding variant pathogenicity using multi-omic pipelines**
Prokisch, Holger (Neuherberg, Germany)

09:55 **The epigenetic clock and metabolism** | *Raj, Kenneth (Cambridge, UK)*

10:30 – 11:00 | Coffee with the JIMD Editors
Gallery Meeting Room 1 | New Formats

10:30 – 11:00 | **Coffee Break**

11:00 – 12:30 | Gene Therapy Clincial Trials
Freiburg | Parallel Session 3A

Chairs: Schulze, Andreas (Toronto, Canada), van Karnebeek, Clara (Amsterdam, Netherlands)

11:00 **Sustained efficacy and safety at week 52 and up to three years in adults with glycogen storage disease type Ia (GSDIa): results from a phase 1/2 clinical trial of DTX401, an AAV8-mediated, liver-directed gene therapy** | *Derks, Terry G. (Groningen, Netherlands)*

11:15 **Metabolic abnormalities in canavan disease and reduction in CNS N-acetyl-L-aspartate in patients receiving systemic AAV9-mediated ASPA gene transfer** | *Eichler, Florian (Boston, MA, USA)*

11:30 **RGX-111 gene therapy for the treatment of severe mucopolysaccharidosis type I (MPS I): Interim analysis of data from the first in human study** | *Wang, Raymond (Orange, USA)*

11:45 **AT845 gene replacement therapy for Late Onset Pompe disease: preliminary safety and efficacy data from FORTIS, a phase I/II open-label clinical study**
Manera, Jordi Diaz (Newcastle upon Tyne, UK)

12:00 **Safety and efficacy of DTX301 in adults with late-onset ornithine transcarbamylase (OTC) deficiency: A Phase 1/2 Trial** | *Harding, Cary O. (Portland, OR, USA)*

12:15 **From academic clinical development to an approved commercial drug administered in multiple highly specialised centres: arsa-cel, a lentiviral haematopoietic stem-cell gene therapy for early-onset metachromatic leukodystrophy (MLD)** | *Fumagalli, Francesca (Milano, Italy)*

11:00 – 12:30 | Urea Cycle Disorders
Jerusalem | Parallel Session 4B

Chairs: Huidekoper, Hidde (Rotterdam, Netherlands), Nakamura, Kimitoshi (Kumamoto, Japan)

11:00 **Neurometabolic impact of liver transplantation in six patients with argininosuccinate lyase deficiency (ASLD)** | *Siri, Barbara (Roma, Italy)*

11:15 **In vivo lentiviral gene therapy for argininosuccinic aciduria** | *Touramanidou, Loukia (London, UK)*

11:30 **Proof of concept of in vivo mRNA therapy in a mouse model of argininosuccinic aciduria** | *Gurung, Sonam (London, UK)*

11:45 **O-GlcNAcylation enhances CPS1 catalytic efficiency for ammonia and promotes ureagenesis** | *Soria, Leandro R. (Pozzuoli, Italy)*

12:00 **In vivo assessment of ureagenesis using stable isotope tracing to monitor disease and treatment efficacy** | *Poms, Martin (Zurich, Switzerland)*

12:15 **Severity-adjusted evaluation of newborn screening on the metabolic disease course in cytosolic Urea Cycle Disorders** | *Zielonka, Matthias (Heidelberg, Germany)*

11:00 – 12:30 | Mitochondrial Disorders
Porto | Parallel Session 3C

Chairs: Christodoulou, John (Melbourne, Australia), Rahman, Shamima (London, UK)

11:00 **Discovery and replication of novel biomarkers for mitochondrial diseases in a cohort of 2,000 individuals** | *Smirnov, Dmitrii (München, Germany)*

11:15 **Unlocking the mitochondrial genome for gene therapy and modelling disease** | *Koppens, Martijn (Utrecht, Netherlands)*

11:30 **Reversal of m.3243A>G related defects to advance drug screening »hits« towards clinical trials** | *Kozicz, Tamas (Rochester, USA)*

11:45 **Inborn disorders of the malate aspartate shuttle lead to disturbed NAD+/NADH redox balance, glycolysis and defective serine biosynthesis** *Broeks, Melissa (Utrecht, Netherlands)*

12:00 **Ryanodine receptor type 3 variants cause acute episodes of rhabdomyolysis related to abnormal calcium homeostasis and impaired autophagy** *de Calbiac, Hortense (Paris, France)*

12:15 **3-methylglutaconic aciduria type II: The molecular mechanism responsible for the mitochondrial dysfunction** | *Yoo, Sukdong (Yangsan-Si, South Korea/ROK)*

11:00 – 12:30 | Disorders of Vitamins and Cofactors
Kyoto | Parallel Session 3D

Chairs: Hannibal, Luciana (Freiburg, Germany), Schwahn, Bernd (Manchester, UK)

11:00 **Oxidative stress and mitochondrial respiration impairment induced by sulfite in rat brain are prevented by the mitochondria-targeted antioxidant XJB-5-131** *Leipnitz, Guilhian (Porto Alegre, Brazil)*

11:15 **Biochemical investigation of SAM mediated allosteric inhibition of MTHFR** *Blomgren, Linnea (Zurich, Switzerland)*

11:30 **Influence of early identification and therapy on long-term outcomes in early-onset MTHFR deficiency** | *Yverneau, Mathilde (Rennes, France)*

11:45 **Improved biochemical profile after hydroxocobalamin dose-escalation in cobalamin C defect** | *Olivieri, Giorgia (Roma, Italy)*

12:00 **Personalized modeling of altered metabolic states inborn errors of cobalamin metabolism** | *Heinken, Almut (Nancy, France)*

12:15 **Novel biochemical and clinical evidence reveals abnormal concentrations of acetylated amino acids in cerebrospinal fluid in acetyl-CoA transporter deficiency** | *Sikic, Katarina (Zagreb, Croatia)*

12:30 – 14:00 | Alumni Café
Gallery Meeting Room 1 | New Formats

12:30 – 14:00 **| Lunch & Poster Viewing**

12:45 – 13:00 | Science Slam: The World of Epigenetics!
Scientific Theatre | New Formats | Hensel, Kai O. (Wuppertal, Germany)

14:00 – 15:30 | Gene and innovative Therapies
Freiburg | Parallel Session 4A

Chairs: Anikster, Yair (Tel Hashomer, Israel), Grünewald, Stephanie (London, UK)

14:00 **Eladocagene exuparvovec gene therapy improves motor development in patients with aromatic L-amino acid decarboxylase deficiency** | *Hwu, Paul Wuh-Liang (Taipei City, Taiwan)*

14:15 **Intracerebral gene therapy in 2 patients with aromatic-L acid decarboxylase (AADC) deficiency** | *Roubertie, Agathe (Montpellier, France)*

14:30 **Hematopoietic stem & progenitor cell gene therapy for Hurler syndrome: interim clinical results and extensive metabolic correction** | *Tucci, Francesca (Milano, Italy)*

14:45 **Novel gene supplementation, genome editing and cellular therapeutic approaches to treat Glutaric Aciduria Type I** | *Bissig, Karl-Dimiter (Durham, USA)*

15:00 **Sustained efficacy of neonatal AAV gene therapy for maple syrup urine disease in mice** | *Pontoizeau, Clément (Paris, France)*

15:15 **Pegzilarginase efficacy in arginase 1 deficiency: results of the PEACE pivotal phase 3 trial** | *Russo, Rossana Sanchez (Atlanta, USA)*

14:00 – 15:30 | Amino Acid Disorders
Jerusalem | Parallel Session 3B

Chairs: Knerr, Ina (Dublin, Ireland), Kozich, Viktor (Praha, Czech Republic)

14:00 **Efficacy and pharmacokinetics of betaine in CBS and cblC deficiency: a cross over randomized controlled trial** | *Imbard, Apolline (Paris, France)*

14:15 **Branched chain ketoacid dehydrogenase kinase (BCKDK) deficiency: a treatable neurodevelopmental disease amenable to newborn screening** *Constante, Juliana R. (Barcelona, Spain)*

14:30 **The impact of liver transplantation on health-related quality of life in intoxication-type inborn errors of metabolism** | *Greco, Benedetta (Rome, Italy)*

14:45 **Nutrigenomics on the fly: a systematic animal model approach to finding dietary treatments for inherited metabolic diseases** | *Johnson, Travis (Clayton, Australia)*

15:00 **Isoleucine to valine substitutions by IARS1 help maintain cellular function during nutritional stress** | *Kok, Gautam (Utrecht, Netherlands)*

15:15 **Genetic correction of ornithine delta-aminotransferase mutation and metabolomic analyses of iPSC lines derived from gyrate atrophy patients** *Hyvonen, Mervi (Helsinki, Finland)*

14:00 – 15:30 | Neurometabolic Disorders
Porto | Parallel Session 4C

Chairs: Desviat, Lourdes (Madrid, Spain), Mochel, Fanny (Paris, France)

14:00 **Identifying the mechanism by which creatine represses expression of AGAT** | *Lee, Alex (Toronto, Canada)*

14:15 **ASS1 deficiency is associated with impaired neuronal differentiation in zebrafish larvae** | *Seidl, Marie J. (Heidelberg, Germany)*

14:30 **Identification of CIn5 as S-depalmitoylase highlights the significance of reversible protein palmitoylation in childhood dementia and Alzheimer's disease** | *Steinfeld, Robert (Zurich, Switzerland)*

14:45 **Cerebrospinal fluid amino acids glycine, serine, and threonine in nonketotic hyperglycinemia** | *Van Hove, Johan (Aurora, CO, USA)*

15:00 **COX11 defects are a novel cause of an infantile-onset mitochondrial encephalopathy** | *Christodoulou, John (Parkville, Australia)*

15:15 **The time has come for newborn screening for pyridoxine-dependent epilepsy** | *Coughlin, Curtis (Aurora, USA)*

14:00 – 15:30 | Novel Disease Insights

Kyoto | Parallel Session 1D

Chairs: Mills, Philippa (London, UK), Rutsch, Frank (Muenster, Germany)

14:00 **ARSK deficiency – a novel subtype of mucopolysaccharidosis** | *Plecko, Barbara (Graz, Austria)*

14:15 **Glutaminase deficiency impairs neuronal function and can be partially rescued with glutamate supplementation** | *Santra, Saikat (Birmingham, UK)*

14:30 **A case of 3-MCC leading to the discovery of a novel neurodevelopmental syndrome caused by bi-allelic loss-of-function variants in RABGAP1** *Oh, Rachel (Toronto, Canada)*

14:45 **Bi-allelic variants in NAE1 cause intellectual disability, ischiopubic hypoplasia, stress-mediated lymphopenia and neurodegeneration** | *Muffels, Irena J. J. (Utrecht, Netherlands)*

15:00 **Infantile superoxide dismutase 1 deficiency syndrome (ISODDES) is an infantile-onset motor neuron disease with impaired glutathione metabolism** | *Park, Julien H. (Münster, Germany)*

15:15 **A previously undescribed combination of juvenile ALS with crystalline retinopathy caused by a de novo mutation in SPTLC2 causing a shift in substrate specificity of Serine Palmitoyl Transferase towards longer chain acyl-CoA's** | *Verloo, Patrick (Belgium)*

15:30 – 18:30 | Networking Activities

19:30 – 00:00 | Networking Evening with Dinner Speech

Konzerthaus | Networking

Hans-Albert Stechl

Lukewarm pumpkin salad

This pumpkin salad with its autumnal flavour gets its culinary appeal from the combination of oriental spices, the sweetness of dates and the bitter-tart freshness of radicchio.

Cut the pumpkin into quarters and scrape out the fibrous inside with the seeds using a spoon. Depending on the type of pumpkin you choose, you may have to peel the squash. Hokkaido pumpkin can spare you this work. It would be a mistake, however, to focus only on making the work easier and leave out many other tasty pumpkin varieties, such as the butternut (nutty flavour) or the chestnut squash (reminiscent of chestnuts).

Cut the pumpkin into cubes of about 2 centimetres. First marinate the pumpkin cubes. For the marinade, finely grate the ginger, finely

crush the cumin in a mortar and mix together with cinnamon and olive oil.

Put the pumpkin cubes in a bowl, pour the marinade over them, mix everything well and leave to marinate for 15 minutes. Spread on a baking tray and cook in an oven preheated to 200 degrees (top and bottom heat, middle shelf). This takes between 20 and 30 minutes, depending on the type of pumpkin. The pumpkin should be soft yet firm, it should never be overcooked or mushy.

Prepare the couscous according to the instructions on the packet. Loosen the couscous well with a fork.

Pit the dates and cut them into small pieces. Pluck the parsley and chop finely. Wash the radicchio, spin dry and cut or pluck into large pieces. Put together in a large bowl with the pumpkin cubes, which should still be warm, and the couscous.

The vinaigrette consists of olive oil, lemon juice, honey, pepper and, to spice it up, add a pinch of chilli powder. Mix everything well and pour over the salad, mix thoroughly and leave to infuse for a few minutes. Arrange the salad on a platter. Sprinkle with a little coarse salt and coarsely ground pepper and finally sprinkle with the finely chopped parsley.

Toasted white bread goes very well with this dish. You can also cube the bread (sugar cube size), fry it in butter in a pan until golden and then incorporate as croutons into the salad.

Ingredients *(for 4 servings)*

1 pumpkin weighing approx. 1 kg

Marinade
1 thumb-sized piece of ginger
1 heaped teaspoon ground Cinnamon
1 heaped teaspoon of ground Cumin seeds
1 dash olive oil

200 g couscous
1 small head of radicchio
10 dates
½ bunch flat-leaf parsley

Vinaigrette
1 tablespoon honey
4 tablespoons olive oil
1 lemon
Salt and pepper
1 pinch of chilli powder
1-2 baguettes
A little butter

For more recipes please visit: www.freiburger-marktkalender.de

08:45 – 10:30 | Epigenetics meets Genetics
Freiburg | Plenary Session 6

Chairs: Baumgartner, Matthias (Zurich, Switzerland), Quelhas, Dulce (Porto, Portugal)

08:45 **Introduction into the session** | *Spiekerkötter, Ute (Freiburg, Germany)*

08:55 **Clinical epigenomics meets metabolic diseases and environment**
Sadikovic, Bekim (London, Canada)

09:25 **Developmental plasticity: the 3rd dimension of phenotypic variation and disease risk** | *Nadeau, Joseph (Scarborough, USA)*

09:55 **Beck-Fahrner syndrome: Delineation of a human mendelian disorder of the DNA demethylation machinery** | *Fahrner, Jill A. (Baltimore, USA)*

10:30 – 11:00 | **Coffee Break**

11:00 – 11:45 | Komrower Lecture
Freiburg

Chair: Salomons, Gajja (Amsterdam, Netherlands)

11:00 **On pathways and blind alleys** | *Plecko, Barbara (Graz, Austria)*

11:45 – 12:45 | Special Awards Session
Freiburg | Special Awards Session

Chairs: Salomons, Gajja (Amsterdam, Netherlands), Schiff, Manuel (Paris, France)

11:45 **Characterization of the 1,5-anhydroglucitol transporter SGLT5 that was found to be mutated in a G6PC3-deficient child with a mild form of neutropenia**
Veiga-da-Cunha, Maria (Bruxelles, Belgium)

12:00 **Tailored amino acid treatment for mitochondrial ARS and QARS1 deficiencies**
Muffels, Irena J.J. (Utrecht, Netherlands)

12:15 **O-GlcNAcylation enhances CPS1 catalytic efficiency for ammonia and promotes ureagenesis** | *Soria, Leandro R. (Pozzuoli, Italy)*

12:30 **Deficient protein glycosylation and compromised functional integrity of PMM2 deficient neurons and brain organoids** | *Shah, Rameen (Rochester, USA)*

12:45 – 13:15 | Awards
Freiburg

INVITED SPEAKERS

David Bick
London, UK

Carolyn Broderick
Sydney, Australia

Jill A. Fahrner
Baltimore, USA

Ronan M. T. Fleming
Galway, Ireland

Mathias Heikenwälder
Heidelberg, Germany

Carla Hollak
Amsterdam, Netherlands

Henrietta Hopkins
London, UK

Martina Huemer
Bregenz, Austria

Tanja Krones
Zurich, Switzerland

Ruth Ley
Tübingen, Germany

Christian Lovis
Geneva, Switzerland

Joseph Nadeau
Scarborough, USA

Holger Prokisch
Neuherberg, Germany

Kenneth Raj
Cambridge, UK

Bekim Sadikovic
London, Canada

Nicolas Sireau
Cambridge, UK

Henna Tyynismaa
Helsinki, Finland

SPECIALS & NEW FORMATS

Community social & health Project — Matrix

Eroica_

Children of the dance class from TV Freiburg-Herdern under the lead of Ivett Csikos have expressed »Eroica«, the 3rd symphony of Ludwig van Beethoven through the form of dance. The name Eroica refers to a heroic symphony and was composed from 1802 to 1803. Today the symphony is considered revolutionary. The children were significantly involved in developing the choreography themselves, in order to foster their independence. Taking responsibility for the whole group and being self-disciplined were of major importance during this project. Everything, starting with the planning and designing this performance was developed under the motto: »Growing together«. Together, the children created something very special, where social background or dancing skills made no difference.

Dancers: Alessa Simon, Caelyn Banks, Jana Schepky, Tim Knochel, Nora Spiekerkötter, Maria Bothe, Leonie Klaus, Anna Breit, Ivett Csikós

Everyone_

The piece is about the »Declaration of human rights« in 1948. To the music of Max Richters "Voices", Matrix highlights that Everyone is responsible for living and fighting for our human rights. We still have a long way to go until these rights are applied to Everyone!

Dancers: Niko Sonner, Pauline Ihme, Nora Spiekerkötter, Maria Bothe, Leonie Klaus, Anna Breit, Ivett Csikós, Jakob Matthaei und Simon Matthaei

The power of humanity — 250 Years Beethoven

Even 250 years after the birth of the »titan« Ludwig van Beethoven, the humanitarian and enlightening ideas of this musical genius are still highly relevant. Beethoven understood the political »fire« of his works. The meaning of the symphony as an expression of the highest form of humanity and the phrase »all men become brothers« is what made the ode a perfect choice for the European anthem. Today, the European anthem stands as a symbol of »unity in diversity«, which is also the fundamental idea of this community dance project called »The Power of Humanity«. The project introduces children, teenagers and young adults from different social backgrounds to classical music. Just as Prometheus brings fire to humankind, Beethoven's music ignites the creative potential of young people. Tolerance, openness and intercultural diversity can be experienced directly in this dance encounter.

Dancers: Niko Sonner, Pauline Ihme, Nora Spiekerkötter, Maria Bothe, Leonie Klaus, Jakob Matthaei, Simon Matthaei and children

Christina Plötze is a dance lecturer at Freiburg University, as well as coach and choreographer of the Showteam Matrix. In 2004 she founded Matrix, which is the official showteam of the German and the Baden Gymnastics Federation. Matrix was 4 times »The Freiburger team of the year«. The team has performed with great success winning three times the gold medal in the world championship of show performances »Gym for Life world challenge«. Matrix represented the German Gymnastics Federation internationally such as in Tokyo, Fiuggi, Seoul, Busan, Dornbirn, Lausanne, Siauliai, Kapstadt, Helsinki, Oslo and Suwon.

Ivett Csikós has been an active member of the Showteam Matrix and Turnverein Freiburg-Herdern, the gymnastics club in the Freiburg district Herdern, since 2011. She is an aerobic trainer and now coaching the dance class for children (aged 9-14 years) from Turnverein Freiburg-Herdern.

Garrod Award Introduction — Lionel & Demian Martin

Two brothers, two instruments. Lionel and Demian Martin, aged 19 and 24, have reunited as a duo 3 years ago. In the meantime, Lionel Martin has become internationally known on the cello and has toured the world as a soloist with well-known orchestras and in a wide variety of chamber music ensembles, including a concert with Anne-Sophie Mutter.

The older brother Demian established himself as a piano cabaret artist and film music composer while studying piano. In sparkling unity they perform their repertoire, ranging from classical sonatas to modern character pieces, enriched by audience-requested improvisation encores.

Behind Human Understanding — Sophie Hauenherm

Introducing the session »Reality meets Metabolism« Sophie Hauenherm is going to present her choreography »Behind human understanding«. She created the dance as her Bachelor thesis at the end of her stage dance studies. Just six months before her finals, she developed an abscess in the region of the thoracic spinal cord and the doctors made the devastating diagnosis of an incomplete paraplegia. Instead of taking the diagnosis as it was, she was facing the challenge. She fought her way back to a life largely independent from the wheelchair. She would not accept her physical constraint and far more important she wouldn't let her dance be defined by it.

Today Sophie Hauenherm is proud of her way and tells her story through her dance. She wants to inspire and encourage other people with her way of dealing with her diagnosis and wants to build awareness.

Science Slam — Dr. Kai Hensel

Dr. Kai Hensel will present a science slam on the topic: The World of Epigenetics in the Scientific Theatre.

A science slam is an increasingly popular format of scientific communication in which the speakers present their research fields to a general (often non-expert) audience. Success is judged by the audience according to the educational content and, importantly, also the entertainment value. Recently, Dr. Hensel has mastered the genre winning multiple championships, finishing as runner-up for Germany (region West) and as national science slam championship finalist.

Dr. Hensel is a German born physician scientist who worked as a consultant in the Department of Pediatrics at Cambridge University Hospital before he moved to Göttingen, Germany. As from October 2022 he will be the Director of the Helios University Children's Hospital Wuppertal and Professor of Clinical Pediatrics at Witten/Herdecke University.

For his PhD he studied epigenetics of virus-host interactions in hepatitis B virus infection. His clinical research interests include advanced cardiovascular imaging techniques, health service sciences and digital healthcare innovations.

Dr. Hensel is also the creator and host of a new clinical pediatrics podcast (currently the most popular podcast on pediatric medicine in German language), called "Die Expertise-Piraten" - available at www.expertise-piraten.eu.

Scientific Theatre

1. Parallel to the poster walk, the 10 high-ranked posters will be presented in the Scientific Theatre.

Key facts

Where:	Foyer
When:	Wed, 31 August, 18:45 – 20:15 h
How:	poster authors present their poster abstract
Who:	all interested participants

2. The patient group representatives will discuss with the audience the patients' needs. A representative from Eurordis will also be present.

Key facts

Where:	Foyer
When:	Wed, 31 August, 13:00 – 13:45 h
Who:	all interested participants

3. Science Slam by Kai Hensel: The World of Epigenetics!

Key facts

Where:	Foyer
When:	Thu, 1 September, 12:45 - 13:00 h
Who:	all interested participants

Speed Mentoring

As a young professional in the field of metabolic medicine you will have the unique opportunity to speak one-on-one with advanced researchers and clinicians within and outside your speciality. During individual interviews of 7 minutes with up to 5 different mentors, you can get insider tips on how to develop a career plan, articulate career transitions and promotions, manage challenging situations at work and more. Bring your questions! This event will allow you to meet with different mentors in a protected professional environment.

Key facts

Where:	Meeting Room 1
When:	Tue, 30 August, 12:45 – 13:45 h Wed, 31 August, 07:30 – 08:30 h Thu, 1 September, 07:30 – 08:30 h Fri, 2 September, 07:30 – 08:30 h
How:	rolling system with different mentors
Who:	clinicians, scientists and laboratory specialists in training under the age of 35
Registration:	online, starting in August

ONLY LIMITED PLACES

Patient Organisations

The SSIEM symposium provides a platform for national and international patient organisations to connect with each other, discuss current challenges, and to join forces. The main topic of this side meeting is to identify «Patients' needs". After this meeting there will be an open discussion in the scientific theatre with everyone interested in the topic.

Key facts

Where:	London
When:	Wed, 31 August, 08:45 – 10:15 h
How:	meeting for patients and patient representatives, international networking, introduction lecture by EURORDIS representative Dr. Gulcin Gumus
Who:	patients and patient representatives
Registration:	patients and patient representatives, open to all interested participants

Alumni Café

For the first time, an Alumni Café will be held at the annual symposium to collect impressions, highlights and developments from the past 50 years. A place where we shed light on the history of the SSIEM as well as the trends in Metabolic Medicine. A place where old friends and colleagues meet again and share their memories in an informal and enjoyable atmosphere.

Key facts

Where:	Gallery Meeting Room 1
When:	Thu, 1 September, 12:30 – 14:00 h
How:	sharing memories of the last 50 years
Who:	retired and emerited colleagues and everybody who likes to connect with them

NETWORKING PROGRAMME

The main aim of our symposium is to share the latest research in the field of metabolic and genetic diseases – but of course it should also be a platform to socialise and network with old and new colleagues and friends.

Welcome Reception

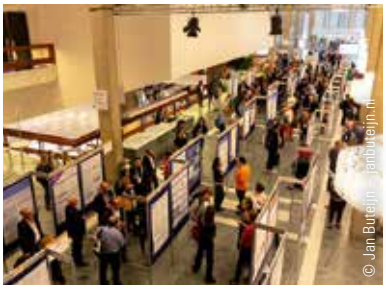
Tue, 30 August, 18:30 – 21:00 h, Foyer

Finally – let us meet again in person for our scientific exchange at the SSIEM 2022! Networking is not least important and often is the motor for new research ideas. To initiate this year's SSIEM symposium we welcome you to Freiburg with a special reception.

Kindly supported by FREI BURG

Poster Walk

Wed, 31 August, 18:45 – 20:15 h, Messe Freiburg



At our Poster Walk you will have the chance to discuss the presented findings with the abstract authors. In parallel, the 10 highest ranked posters by young investigators will be presented in 7-minute contributions at our scientific theatre and there will be a floor for an open discussion.

Young SSIEM Evening

Wed, 31 August, 20:30 – 00:00 h, Freiburg City Centre

Young colleagues from all professions within the SSIEM (and of course those who like to connect with the young) are invited to better get to know each other and exchange ideas at this special Young SSIEM Evening. There is no age limit and everybody is welcome – but the idea of this special networking event is to strengthen the young SSIEM!

Registration: online – 15 EUR

Networking Evening

Thu, 1 September, 19:30 – 00:00 h, Konzerthaus Freiburg

Connect with participants from all over the world in a great atmosphere. We will bring the Black Forest into the heart of Freiburg and will give you a feeling of this special forest region in the southwest of Germany. It will be a special night and not at least a floor for personal exchange.

Registration: online – 30 EUR

GENERAL INFORMATION

Date
30 August – 2 September 2022

Website
www.ssiem2022.org

Symposium Venue
Messe Freiburg
Neuer Messplatz 1
79108 Freiburg, Germany

Symposium President
Prof. Dr. Ute Spiekerkötter
University Hospital of Freiburg
Freiburg, Germany

Congress Management
Intercongress GmbH
Alisa Ganter
Ingeborg-Krummer-Schroth-Str. 30
79106 Freiburg, Germany
ssiem@intercongress.de
www.intercongress.de

Exhibition Management and Sponsoring
Society for the Study of Inborn Errors
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Ralph Kerschbaumer – Corporate Liaison Officer
PO Box 3375, South Croydon, CR2 1PN
United Kingdom
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corporateliaison@ssiem.org
www.ssiem.org

Symposium Office

Monday, 11:00 – 18:00 h

Tuesday, 07:30 – 18:30 h

Wednesday, 07:00 – 19:00 h

Thursday, 07:00 – 15:30 h

Friday, 07:00 – 13:00 h

Media Check for Speakers

Monday, 13:30 – 16:30 h

Tuesday, 07:30 – 19:00 h

Wednesday, 06:30 – 19:00 h

Thursday, 06:30 – 15:30 h

Friday, 07:30 – 13:00 h

WiFi Access

Network: SSIEM2022

Password: SSIEM2022

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CME Certification

An application has been made

to the UEMS EACCME®

for CME accreditation of this event.

Registration fees

SYMPOSIUM TICKET 4 DAYS	LATE REGISTRATION FROM 7 JULY 2022
SSIEM Member	550 EUR
Non-member	800 EUR
Students/Residents-in-Training (<35 yrs)	400 EUR
Dieticians & Nurses (reduced fee)	400 EUR

SINGLE DAY TICKET 1 DAY	LATE REGISTRATION FROM 7 JULY 2022
SSIEM Member	200 EUR
Non-member	300 EUR
Dieticians & Nurses (reduced fee)	200 EUR

Please register online at **www.ssiem2022.org/registration**

REGISTRATION FEE INCLUDES

Full Delegate (SSIEM Members, non-members, students & residents-in-training):

- Admission to all oral and poster sessions and exhibition area
- Scientific programme and symposium bag
- Access to the online version of the abstract book
- Coffee, tea and lunch bag during symposium breaks
- Welcome Reception
- Free public transport within Freiburg from 29 August to 2 September 2022

Networking Evening and Social Programme are to be paid for extra.

SINGLE DAY TICKET

- Admission to all oral and poster sessions and exhibition area
- Scientific programme and symposium bag
- Access to the online version of the abstract book
- Coffee, tea and lunch bag during symposium breaks of the day/ days of the ticket
- Free public transport within Freiburg on the day/ days of the ticket

Single day tickets do not allow access to social events. No more than two consecutive single day tickets can be purchased by one delegate.

Alterations & cancellation of congress registration

Written notification is required for all congress cancellations and name changes. Notifications should be sent to the SSIEM 2022 congress office **ssiem@intercongress.de**

For alterations of reservations or any other invoiced item an administrative fee of 20 EUR will be charged. A cancellation fee of 50 EUR will be charged for cancellations until 2 August 2022. The fee for congress participation will not be refunded, even in case of non-participation, if a cancellation is not received in time. A refused visa application is no reason for refund. Alternative participants are readily accepted for an administrative fee of 20 EUR. No reimbursement will be made in case of cancellation of a social programme from a participant. Please find full terms and conditions online at **www.ssiem2022.org/registration**

SSIEM

Become a member of SSIEM | To benefit from a reduced registration fee, apply to become a member of SSIEM today. Simply visit our website – **www.ssiem.org** – to see how.

Messe Freiburg © Florian Bilger Fotodesign

SSIEM APP

GET THE SSIEM MOBILE APP!

- Access the scientific programme, networking programme, satellite symposia and faculty list
- Read the abstract texts
- Create your own daily schedule
- Visit partner and exhibitor profiles
- Stay informed and receive the latest news
- Chat with other attendees
- Post on our social feed

HOW TO DOWNLOAD:

The easiest way to download the mobile app is to scan this code. You can also search for **SSIEM** in the Apple Store or Google Play Store. Once you have installed the SSIEM app, you can access the SSIEM 2022 Congress by clicking on the top tile. After downloading the app, do not forget to enable push notifications to stay up-to-date on the latest news!

HOTEL

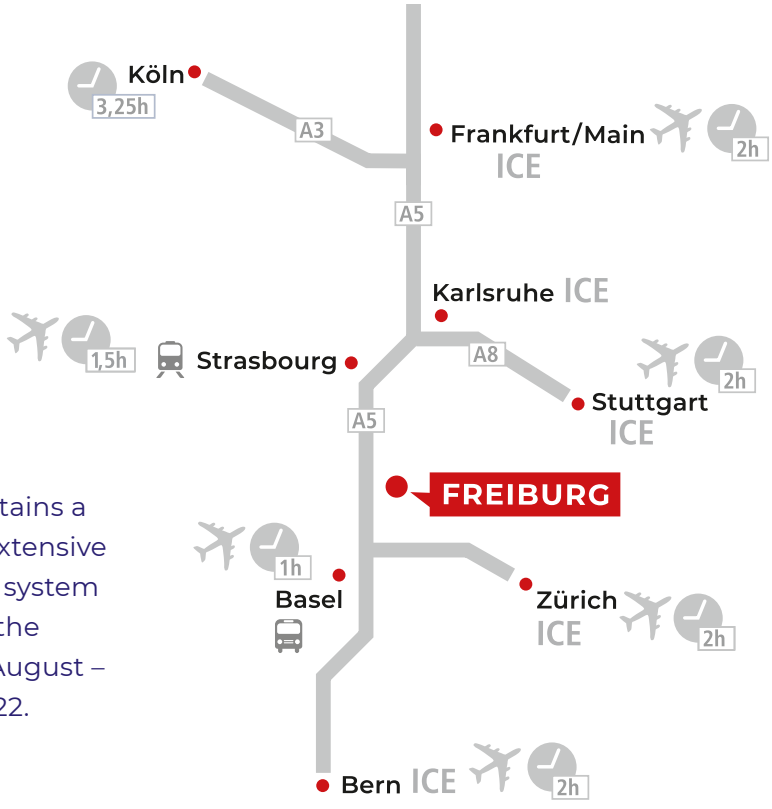
The Freiburg Convention Bureau has composed a list of many hotels in Freiburg, which you can find and book online at **www.ssiem2022.org/hotel**

For further information or group bookings please contact Kerstin Schultheis from Freiburg Convention Bureau via **groups@fwtm.de** or phone **+49 761 3881 1516**

TRAVEL

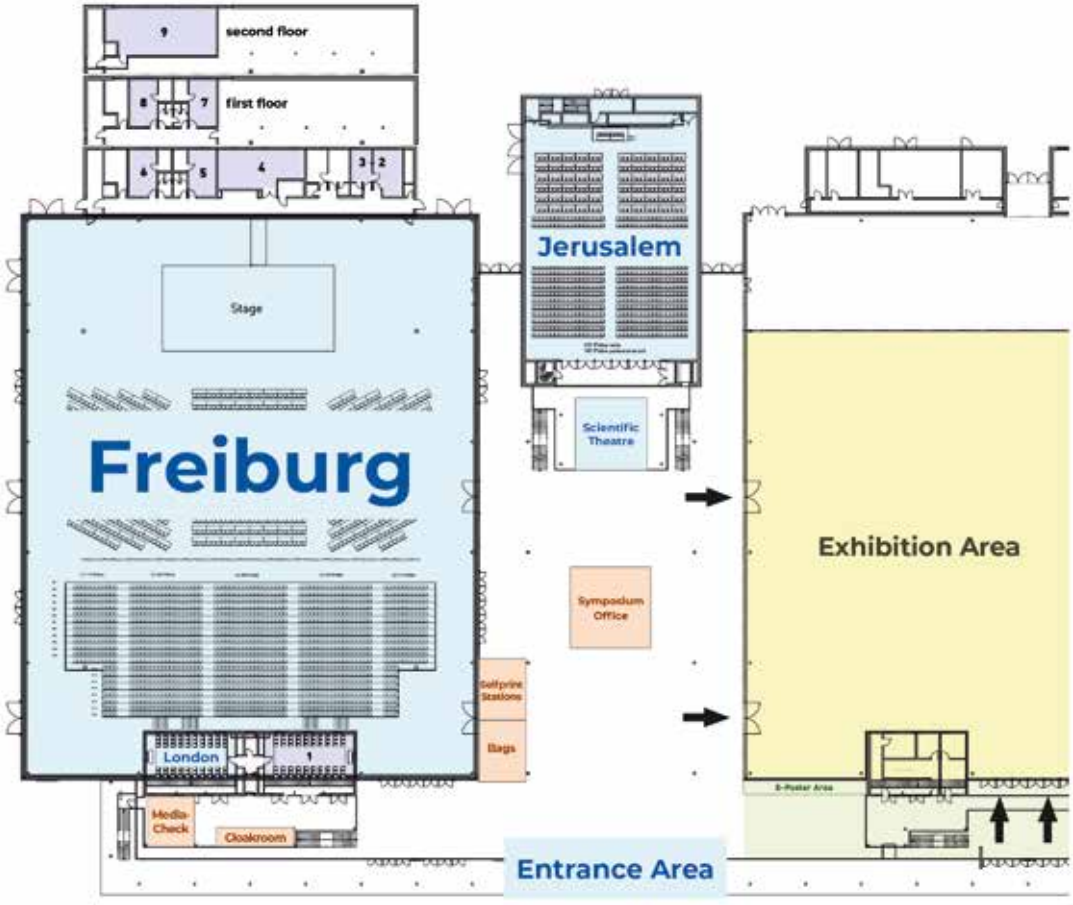
The Messe Freiburg is easily reachable whether you are traveling by air, car, train or bus.★

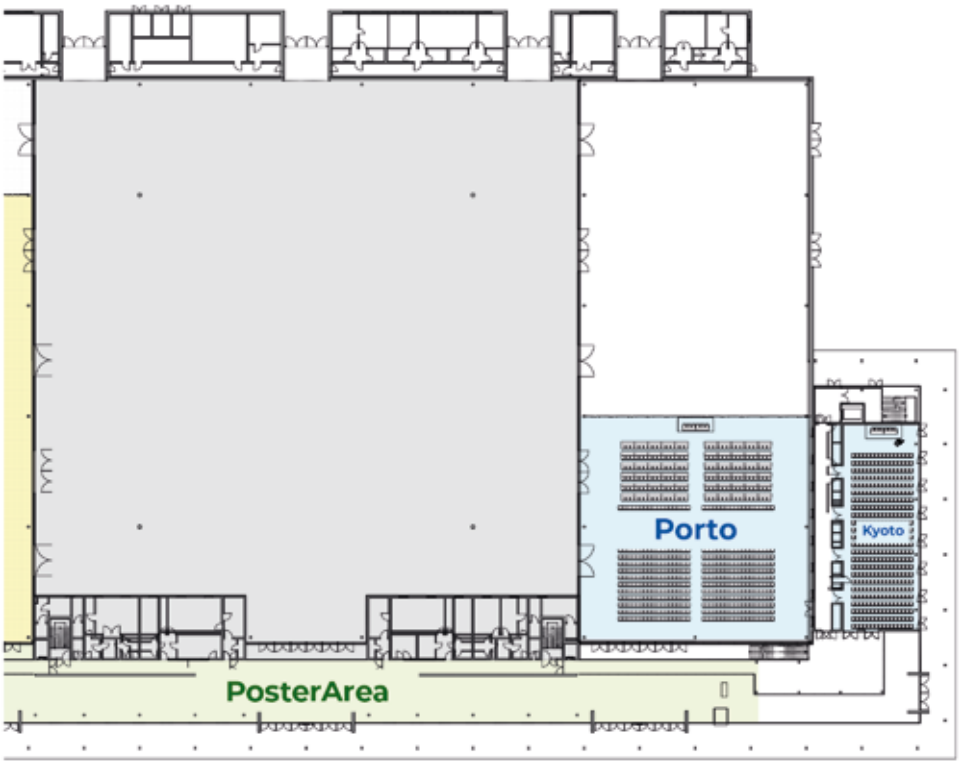
For more information about the best way to travel to Freiburg and for special rates please visit our website **www.ssiem2022.org/travel**



★ Your eTicket contains a free use of the extensive public transport system in Freiburg and the region from 29 August – 2 September 2022.

- Conference facilities
- Services
- Poster area
- Industry / exhibition area
- Meeting rooms

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SSIEM Annual Symposium 2023
29 August - 1 September 2023
Jerusalem, Israel
החברה הישראלית למחלות מטבוליות
Israel Society for Metabolic Diseases
Important Dates & Deadlines
Start Online Registration: 4th January 2023
Start Call for Abstracts: 16th January 2023
Abstract Submission Deadline: 19th April 2023
ssiem2023.org

Freiburg
IM BREISGAU

Freiburg is located in the lower Rhine Valley between the mountain ranges of the Vosges and the Black Forest in the border-triangle of Germany, Switzerland and France. It is not only known as the »Green City« and »Solar City« of Germany, but is also the sunniest and warmest city in Germany. Freiburg was founded in the early 12th century. Nowadays, it has about 230,000 inhabitants. With its famous medieval old town, its modern buildings and its surrounding vineyards, unspoiled forests and rolling hills it combines urban lifestyle with environment in a special way like no other city.


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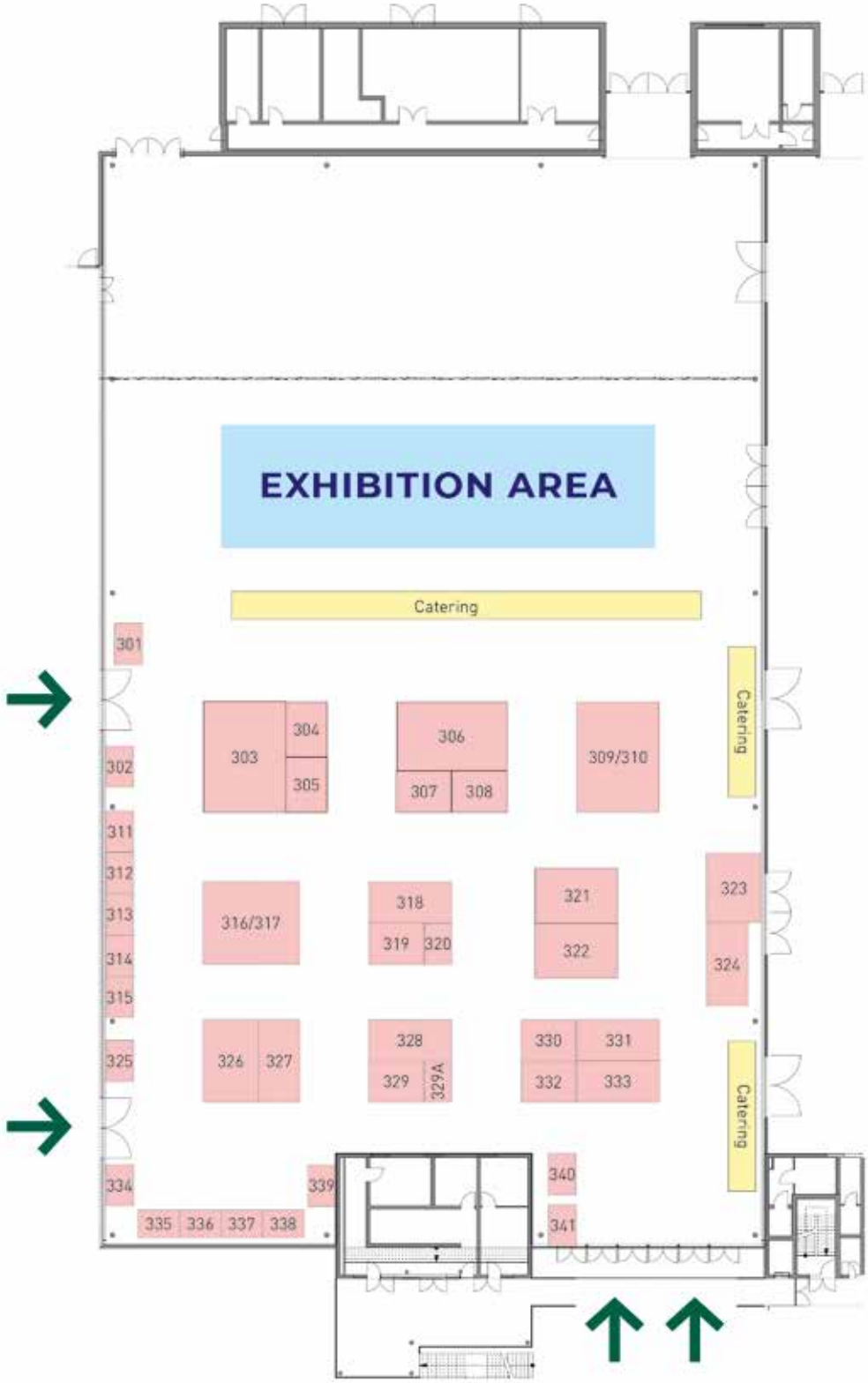
EXHIBITION AREA



EXHIBITORS

339	Aceragen
330	Aeglea BioTherapeutics
301	Ajinomoto Cambrooke
341	Alfasgima S.P.A.
316	Amicus Therapeutics
317	Amicus Therapeutics
323	APR Applied Pharma Research s.a.
322	ARCHIMEDlife/ amedes
327	Astellas Gene Therapies
314	Baebies
306	BioMarin
315	Bruker BioSpin
304	CENTOGENE
303	Chiesi Farmaceutici spa
334	CTI Clincial Trial & Consulting
336	Denali Therapeutics
319	Dipharma SA
340	Egetis Therapeutics
338	Galen
311	GC Biopharma co.
332	Immedica Pharma
325	JCR Pharmaceuticals
335	Labsystems Diagnostics
312	Lactalis Nutrition Sante
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324	Nutricia
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SATELLITE SYMPOSIA & SPONSORING

INVITATION

SSIEM 2022 Satellite Symposia sponsored by BioMarin

Creating waves with PALYNZIQ®▼ (pegvaliase): Three years of experience in Europe

Tuesday 30 August 2022 | 12:45-13:45 hrs, Hall Freiburg, Messe Freiburg, Germany

Moderator

Cary Harding, MD
Oregon Health & Science University, Portland, OR, USA

Experience with PALYNZIQ® in the clinic

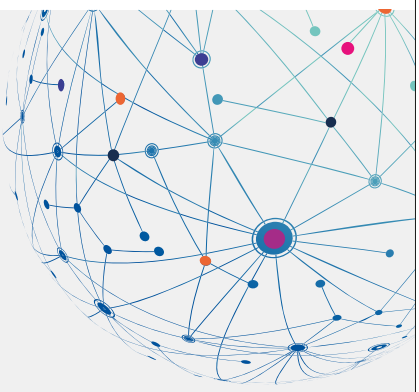
Ania Muntau, MD
University Children's Hospital, University Medical Center Hamburg-Eppendorf, Hamburg, Germany

Valentina Rovelli, MD
ASST Santi Paolo e Carlo, San Paolo Hospital, University of Milan, Milan, Italy

▼This medicinal product is subject to additional monitoring. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse reactions. Healthcare professionals should report adverse events in accordance with their local requirements. Adverse events should also be reported to BioMarin on + 1 415 506 6179 or drugsafety@bmrn.com.

PALYNZIQ® Abbreviated Prescribing Information

Improving the diagnostic pathway in MPS and treating Morquio A in the real world: best practice and findings from MARS



Thursday 1 September 2022
12:45-13:45 hrs, Hall Freiburg, Messe Freiburg, Germany

Dr. Ekkehart Lausch University of Freiburg, Germany
Dr. Karolina Stepien Salford Royal Hospital, Manchester, UK
Prof. Fatih Ezgü Gazi University, Ankara, Turkey

BIOMARIN

These symposia have been initiated, organised and funded by BioMarin. These symposia are intended for healthcare professionals registered for SSIEM 2022 and will include information about licensed BioMarin products.

EU-MPRL-00044
July 2022

| **TUESDAY** | **30 AUGUST 2022** | HALL FREIBURG | 12.45-13.45 |

Creating waves with PALYNZIQ®▼ (pegvaliase): Three years of experience in Europe

Chair: Cary Harding, Portland, United States

Welcome & introduction: PALYNZIQ®▼ three years on

Cary Harding, Portland, United States

Riding the waves of change: My experience with PALYNZIQ®▼

Valentina Rovelli, Milan, Italy

My journey with PALYNZIQ®▼: A smooth sea does not make a skilled sailor

Ania Muntau, Hamburg, Germany

Q & A

Cary Harding, Portland, United States

sponsored by BIOMARIN®

NOTES

| **TUESDAY** | **30 AUGUST 2022** | HALL JERUSALEM | 12.45-13.45 |

Optimization of long-term management of patients with organic acidaemia (propionic and methylmalonic acidurias)

Chairs: Sufin Yap, Sheffield, United Kingdom and Frank Rutsch, Münster, Germany

Introduction & Disease Overview

Sufin Yap, Sheffield, United Kingdom and Frank Rutsch, Münster, Germany

The metabolism and biochemistry we need to unravel
Urea and Krebs Cycle interplay & impairment in PA & MMA

François Feillet, Nancy, France

Real-world evidence with Carbaglu®

The Prospective Observational study of long-TErm Carbaglu® for the Treatment of PA & MMA study (PROTECT study)

Understanding the long-term Management of Organic Acidemia Patients with Carbaglu®: A Mixed Methods Approach

Sufin Yap, Sheffield, United Kingdom

Long-term management & treatment of patients with propionic and methylmalonic acidurias with Carbaglu® — Experience in UK, France & Germany

Sufin Yap, Sheffield, United Kingdom,
François Feillet, Nancy, France,
Clemens Kamrath, Giessen, Germany

Three-year follow-up of patients with propionic and methylmalonic acidurias Long-term Carbaglu® vs liver transplant

Shirou Matsumoto, Kumamoto, Japan

Q&A and Closing Remarks

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Focused on the Few

A Medscape LIVE! EVENT

CPD

Clinician and Patient Perspectives in the Management of Long-Chain Fatty Acid Oxidation Disorders

TUESDAY, 30 AUGUST, 2022 | 12:45 – 13:45 CET

REGISTRATION: 12:30 CET | **PRESENTATION:** 12:45 CET

MESSE FREIBURG, NEUER MESSPLATZ 1

79108 FREIBURG, GERMANY | **ROOM:** HALL PORTO

IN-PERSON EVENT

MODERATOR

Jerry Vockley, MD, PhD
Cleveland Family Endowed Chair in Pediatric Research
Professor of Human Genetics
University of Pittsburgh
Chief of Genetic and Genomic Medicine
Director of the Center for Rare Disease Therapy
UPMC Children's Hospital of Pittsburgh
Pittsburgh, Pennsylvania, United States

PANELISTS

Daniela Karall, MD, IBCLC
Professor of Pediatrics
Inherited Metabolic Disorders
Clinic for Pediatrics
Medical University of Innsbruck
Innsbruck, Austria

Stephanie Grünewald, MD, PhD
Consultant in Metabolic Medicine
Great Ormond Street Hospital for Children NHS Foundation Trust
Senior Lecturer
Institute of Child Health
London, United Kingdom

For more information and to register, visit:
www.medscape.org/symposium/LC-FAOD-2022

Supported by an independent educational grant from ultragenyx

Medscape EDUCATION GLOBAL

| **TUESDAY** | **30 AUGUST 2022** | HALL PORTO | 12.45-13.45 |

Clinician and Patient Perspectives in the Management of Long-Chain Fatty Acid Oxidation Disorders

Chair: Jerry Vockley, Pittsburgh, United States

Welcome and Introductions

Jerry Vockley, Pittsburgh, United States

Recognizing LC-FAODs Through Clinician and Patient Eyes

Stephanie Grünewald, London, United Kingdom

The Holistic Management of LC-FAOD: Combination Strategies

Daniela Karall, Innsbruck, Austria

Concluding Remarks

Jerry Vockley, Pittsburgh, United States

Audience Q & A

sponsored by ultragenyx

The FACEs* of Fabry disease

*FACEs: Fabry Associated Clinical Events – renal, cardiac and cerebrovascular outcomes

Welcome and introductions: Ana Jovanovic, Salford, United Kingdom

Patient perspectives: Experiences with FACEs

- Two people living with Fabry disease share their experiences of FACEs in a tructured interview led by Ana Jovanovic
- Personal impact of FACEs on the people living with Fabry disease and their families

People living with Fabry disease and Ana Jovanovic, Salford, United Kingdom

Session 1: Overview of FACEs

Presentation of an overview of FACEs, including:

- The multisystemic nature of Fabry disease and preclinical manifestations
- An introduction to FACEs, including use as a treatment outcome (not just as a clinical trial tool) and impact on clinical management
- The effect of timely treatment in Fabry disease and the potential impact on FACEs (including morbidity and mortality)
- Cardiac FACEs – impact on QoL and correlation with other measures, based on clinical experience

Peter Nordbeck, Würzburg, Germany

Session 2: Incidence of FACEs during migalastat treatment

Presentation of FACE incidence in patients receiving migalastat, including:

- Overview of previous data in the literature surrounding FACE incidence
- Incidence of FACEs during a long-term post-hoc integrated analysis:
  - Overall incidence and incidence by event category (renal, cardiac and cerebrovascular)
  - Factors affecting time to first FACE
  - Association between baseline variables and rate of FACEs (highlighting the importance of timely treatment initiation to manage disease)
- followME baseline data – comparative history of FACEs and comorbidities across arms

Gere Sunder-Plassmann, Vienna, Austria

Live Q & A

Moderated by Ana Jovanovic with Peter Nordbeck and Gere Sunder-Plassmann

sponsored by Amicus Therapeutics

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Satellite Symposia

Our enduring commitment to people living with *rare diseases*

Come visit us at our booth

Satellite Symposia INVITATION

August 31st, 2022, 7:30 AM - 8:30 AM
Olipudase alfa: A New Treatment For Patients with ASMD

Chair: Dr. Eugen Mengel, MD
SphinCS GmbH, Hochheim, Germany

Dr. Nathalie Guffon, MD
Hôpital Femme Mère Enfant, Bron, France

Professor Maurizio Scarpa, MD, PhD
Udine University Hospital, Udine, Italy

September 1st, 2022, 7:30 AM – 8:30 AM
Unmet Needs in Fabry Disease: Where To Go From Here?

Chair: Dr. Christoph Wanner, MD
University Hospital of Würzburg, Germany

Dr. James Moon, MD, MRCP, MB BCh
University College London and Barts Heart Centre- UK

Dr. Fatih Ezgü, MD
Gazi University Hospital, Ankara, Turkey

September 1st, 2022, 12:45 PM-1:45 PM
Pompe Disease, A New Era: The Latest avalglucosidase alfa Clinical Data and Early Real-World Experiences

Chair: Prof. Andreas Hahn, MD
Department of Child Neurology, University Hospital - Giesen, Germany

Prof. Priya Kishnani, MD, MB BS
Duke University School of medicine, Durham, North Carolina, USA

Dr. Serena Gasperini
San Gerardo Hospital,
University of Milano Bicocca, Italy

Sanofi-Aventis Deutschland GmbH - Lützowstraße 107
D-10785 Berlin – Germany
Approval Date: July 2022
MAT-GLB-2203155 v1.0

| **WEDNESDAY** | **31 AUGUST 2022** | HALL PORTO | 07.30-08.30 |

Olipudase alfa: a new treatment for patients with ASMD

Chair: Eugen Mengel, Hochheim, Germany

Welcome and introduction

Eugen Mengel, Hochheim, Germany

Sphingomyelin accumulation — Pathology behind the disease: sphingomyelin accumulation and clinical manifestations

Nathalie Guffon, Bron, France

Olipudase alfa data — Investigating olipudase alfa in clinical trials: efficacy and safety

Maurizio Scarpa, Udine, Italy

ASMD/olipudase alfa clinical case — The treatment outcome: an ASMD patient case study

Eugen Mengel, Hochheim, Germany

Panel discussion and Q&A

All panel

Closing remarks Eugen Mengel, Hochheim, Germany

sponsored by sanofi

TRAVERE™ THERAPEUTICS

Classical Homocystinuria and Development of a New Enzyme Replacement Therapy

Wednesday, 31 August 2022, 07:30–08:30 CEST
SY6 Satellite Symposium, Kyoto meeting room

Breakfast will be provided

Travere looks forward to welcoming you to a satellite symposium at SSIEM 2022.

We hope you can join our renowned experts **Dr Can Ficicioglu** and **Dr Andrew Morris** to discuss current and future management of classical homocystinuria (HCU)

Agenda

Welcome and Introduction
Can Ficicioglu & Andrew Morris

Overview of Classical HCU
Andrew Morris

Current Management of Classical HCU
Andrew Morris

Case Study: Patient Journey and Experience with Classical HCU
Can Ficicioglu

COMPOSE Phase 1/2 Trial: Overview of Interim Data on Pegtibatinase
Can Ficicioglu

Your Questions Answered
Can Ficicioglu & Andrew Morris

Speakers

Can Ficicioglu
MD, PhD

Professor of Pediatrics,
University of Pennsylvania, USA

Director, Newborn Screening &
Lysosomal Storage Disease Programs,
Children's Hospital of Philadelphia

Andrew Morris
MBBS, MD, MRCP

Consultant Clinical Paediatrician in
Inherited Metabolic Medicine
University of Manchester, UK

This meeting is open to all registered delegates at the SSIEM 2022 congress.

| **WEDNESDAY** | **31 AUGUST 2022** | HALL KYOTO | 07.30-08.30 |

Classical Homocystinuria and Development of a New Enzyme Replacement Therapy

Welcome and Introduction
Can Ficicioglu, Philadelphia, United States
Andrew Morris, Manchester, United Kingdom

Overview of Classical HCU
Andrew Morris, Manchester, United Kingdom

Current Management of Classical HCU
Andrew Morris, Manchester, United Kingdom

Case Study: Patient Journey and Experience with Classical HCU
Can Ficicioglu, Philadelphia, United States

COMPOSE Phase 1/2 Trial: Overview of Interim Data on Pegtibatinase
Can Ficicioglu, Philadelphia, United States

Your Questions Answered
Can Ficicioglu, Philadelphia, United States
Andrew Morris, Manchester, United Kingdom

sponsored by

TRAVERE™ THERAPEUTICS

| **WEDNESDAY** | **31 AUGUST 2022** | HALL JERUSALEM | 18.45-19.45 |

Reaching new hights in ex vivo gene therapy — therapeutic advances for early onset MLD patients

Chair: Simon Jones, Manchester, United Kingdom

Interactive panel discussion

MLD at a glance

Simon Jones, Manchester, United Kingdom

Libmeldy clinical data

Francesca Fumagalli, Milan, Italy

Gene therapy treatment process — collaboration between MLD and HSCT experts

Caroline Sevin, Paris, France

Peter Van Hasselet, Utrecht, The Netherlands

MLD Newborn screening pilots in Europe

David Kasper, Vienna, Austria

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Orchard therapeutics™

| **WEDNESDAY** | **31 AUGUST 2022** | HALL PORTO | 18.45-20.15 |

Immunogenicity and Investigational AAV Gene Therapy

Chair: Benedikt Schoser, Munich, Germany

Welcome and Introductions

Benedikt Schoser, Munich, Germany

Immune Response: The Basics

Benedikt Schoser, Munich, Germany

Potential Immunogenic Responses to Investigational AAV Gene Therapy

Federico Mingozzi, Spark Therapeutics, Inc., United States

Approaches Under Investigation that May Mitigate Immunogenicity in Investigational AAV Gene Therapy

Antonio Toscano, Messina, Italy

Live Panel Q & A

All faculty

sponsored by

Spark™ THERAPEUTICS

SYMPOSIUM

Transformative science in action:
Gene therapy for AADC deficiency

PTC Therapeutics symposium at the 15th SSIEM Annual Congress

1 September 2022, 07:30–08:30 CEST, Hall Jerusalem, Messe Freiburg, Freiburg, Germany

Time (CEST)	Topic	Speaker
07:30–07:33	Welcome, introductions and objectives	Prof. Volker Mall
07:33–07:48	The eladocagene exuparvovec* clinical programme: The journey so far	Prof. Wuh-Liang Hwu
07:48–08:03	Navigating the AADC deficiency diagnostic journey from the caregiver's perspective	Mr Richard Poulin and Mrs Judy Poulin
08:03–08:18	Real-world eladocagene exuparvovec treatment experience from Montpellier	Prof. Agathe Roubertie
08:18–08:28	Panel discussion	All faculty
08:28–08:30	Summary and close	Prof. Volker Mall

*Eladocagene exuparvovec is indicated for the treatment of patients aged 18 months and older with a clinical, molecular, and genetically confirmed diagnosis of aromatic L-amino acid decarboxylase (AADC) deficiency with a severe phenotype in the European Member States, Iceland, Liechtenstein, Norway, and Northern Ireland (Upstaza Summary of Product Characteristics [SmPC]).

AADC, aromatic L-amino acid decarboxylase; SSIEM, Society for the Study of Inborn Errors of Metabolism.

PTC THERAPEUTICS™

Symposium organised and funded by PTC Therapeutics and intended for healthcare professionals only.

MED-ALL-AADC-2200166
July 2022

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL JERUSALEM | 07.30-08.30 |

Transformative science in action: Gene therapy for AADC deficiency

Welcome, introductions and objectives
Volker Mall, Munich, Germany

The eladocagene exuparvovec clinical programme: The journey so far
Wuh-Liang Hwu, Taipei, Taiwan

Navigating the AADC deficiency diagnostic journey from the caregiver's perspective
Mr Richard Poulin and Mrs Judy Poulin

Real-world eladocagene exuparvovec treatment experience from Montpellier
Agathe Roubertie, Montpellier, France

Panel discussion
All faculty

Summary and close
Volker Mall, Munich, Germany

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PTC THERAPEUTICS

NOTES

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL PORTO | 07.30-08.30 |

Unmet Needs in Fabry disease: Where to go from here?

Chair: Christoph Wanner, Würzburg, Germany

Fabry Disease in 2022: How are we doing?

Christoph Wanner, Würzburg, Germany

Left Ventricular Involvement in Fabry Disease: Consequences and Assessment

James Moon, London, United Kingdom

Life with Fabry Disease: Impact of Pain and GI Disturbances

Fatih Ezgü, Ankara, Turkey

Discussion

Christoph Wanner, Würzburg, Germany
James Moon, London, United Kingdom
Fatih Ezgü, Ankara, Turkey

sponsored by

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL KYOTO | 07.30-08.30 |

Pompe Disease: A Metabolic Disease Impacting Muscle and Potential Muscle-directed Gene Therapeutic Approaches

Challenges in the Diagnosis of Pompe Disease

Tahseen Mozaffar, Orange, United States

Therapeutic Landscape for Pompe Disease and Gene Therapy Approaches

Benedikt Schoser, Munich, Germany

Q & A / Discussion

Moderated by Angela Smith, San Francisco, United States

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL FREIBURG | 12.45-13.45 |

Improving the diagnostic pathway in MPS and treating Morquio A in the real world: best practice and findings from MARS

Chair: Ekkehart Lausch, Freiburg, Germany

Welcome to the meeting

Ekkehart Lausch, Freiburg, Germany

Diagnostic challenges in MPS: can we reduce the diagnostic odyssey?

Fatih Ezgü, Ankara, Turkey

Long-term follow-up of Morquio A: 6-year data from the MARS registry

Karolina Stepien, Manchester, United Kingdom

Q & A panel discussion

Moderator: Ekkehart Lausch, Freiburg, Germany

Speakers: Karolina Stepien, Manchester, United Kingdom

Fatih Ezgü, Ankara, Turkey

Summary and meeting close

Ekkehart Lausch, Freiburg, Germany

sponsored by BIOMARIN

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL JERUSALEM | 12.45-13.45 |

Pompe disease, a new era: the latest avalglucosidase alfa clinical data and early real-world experiences

Chair: Andreas Hahn, Giesen, Germany

Introduction by Chair

Andreas Hahn, Giesen, Germany

COMET, Mini-COMET, and real-world evidence on avalglucosidase alfa in the United States: What data do we have so far?

Priya Kishnani, Durham, United States

Latest perspectives: A case report of switching from alglucosidase alfa to avalglucosidase alfa

Serena Gasperini, Milan, Italy

Looking to the future: Avalglucosidase alfa in infants with Pompe disease (Baby-COMET) and impact of home infusion therapy on quality of life

Andreas Hahn, Giesen, Germany

Panel discussion and Q & A

Andreas Hahn, Giesen, Germany
Priya Kishnani, Durham, United States
Serena Gasperini, Milan, Italy

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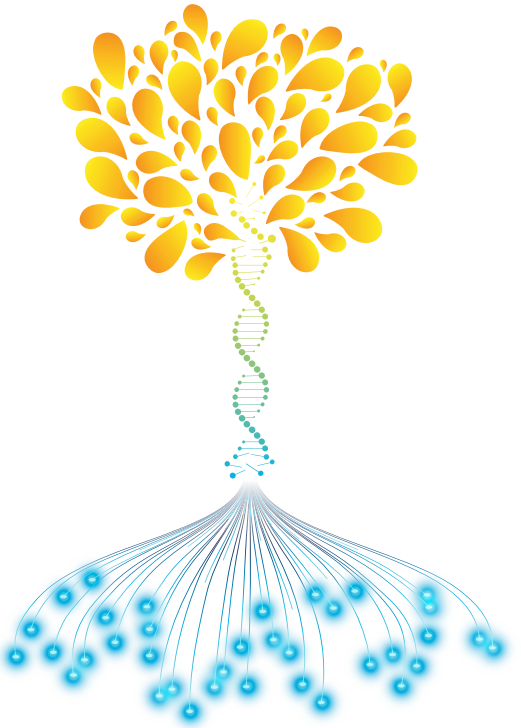
SSIEM ANNUAL SYMPOSIUM

Optimizing **early diagnosis** in lysosomal storage diseases and **innovation for the future**



1 September 2022
12:45–13:45 CEST

A Takeda-sponsored satellite symposium at the Society for the Study of Inborn Errors of Metabolism (SSIEM) 2022 Annual Symposium

Chaired by Prof. Julia Hennermann, this 60-minute symposium aims to explore the innovative advancements for improving early diagnosis of lysosomal storage diseases (LSDs). During four presentations, our expert faculty will draw on their experience to highlight the importance of prompt diagnosis and to discuss thought-provoking developments in shortening the path to rare disease diagnosis through newborn genetic screening and digital technologies.

Presentations

The importance of early diagnosis in Fabry disease and potential innovative approaches

Screen4Care: shortening the path to rare disease diagnosis using newborn genetic screening and digital technologies

Early diagnosis of leukodystrophies: recognizing key signs and symptoms

Newborn screening with a focus on mucopolysaccharidoses (MPS)

Prof. Julia Hennermann (Chair)
University Medical Center Mainz, Mainz, Germany

Dr Edith (Sky) Gross
EURORDIS-Rare Diseases Europe, Paris, France

Prof. Ingeborg Krägeloh-Mann
University Children's Hospital Tübingen, Tübingen, Germany

Prof. Shuan-Pei Lin
Mackay Memorial Hospital, Taipei, Taiwan

This meeting is intended for healthcare professionals only and is initiated, organized and funded by Takeda.

VV-MEDMAT-71248 Date of preparation: July 2022

Takeda

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL PORTO | 12.45-13.45 |

Optimizing early diagnosis in lysosomal storage diseases and innovation for the future

Chair: Julia Hennermann, Mainz, Germany

The importance of early diagnosis in Fabry disease and potential innovative approaches

Julia Hennermann, Mainz, Germany

Screen4Care: shortening the path to rare disease diagnosis using newborn genetic screening and digital technologies

Edith (Sky) Gross, Paris, France

Early diagnosis of leukodystrophies: recognizing key signs and symptoms

Ingeborg Krägeloh-Mann, Tübingen, Germany

Newborn screening with a focus on mucopolysaccharidoses (MPS)

Shuan-Pei Lin, Taipei, Taiwan

sponsored by

Takeda

| **THURSDAY** | **1 SEPTEMBER 2022** | HALL KYOTO | 12.45-13.45 |

What matters most?
Improving care in Lysosomal Storage Disorders (LSDs)

Chair: Maurizio Scarpa, Udine, Italy

Welcome and introductions

Maurizio Scarpa, Udine, Italy

Multidisciplinary care of patients with LSDs

Patricio Ricardo da Terra Aguiar, Lisbon, Portugal

Putting the Fabry disease patient at the heart of management

Uma Ramaswami, London, United Kingdom

Alpha-mannosidosis: where can we make a difference with Enzyme Replacement Therapy (ERT)?

Nathalie Guffon, Lyon, France

20 years of ERT: learnings and aspirations

Maurizio Scarpa, Udine, Italy

Q & A

All faculty

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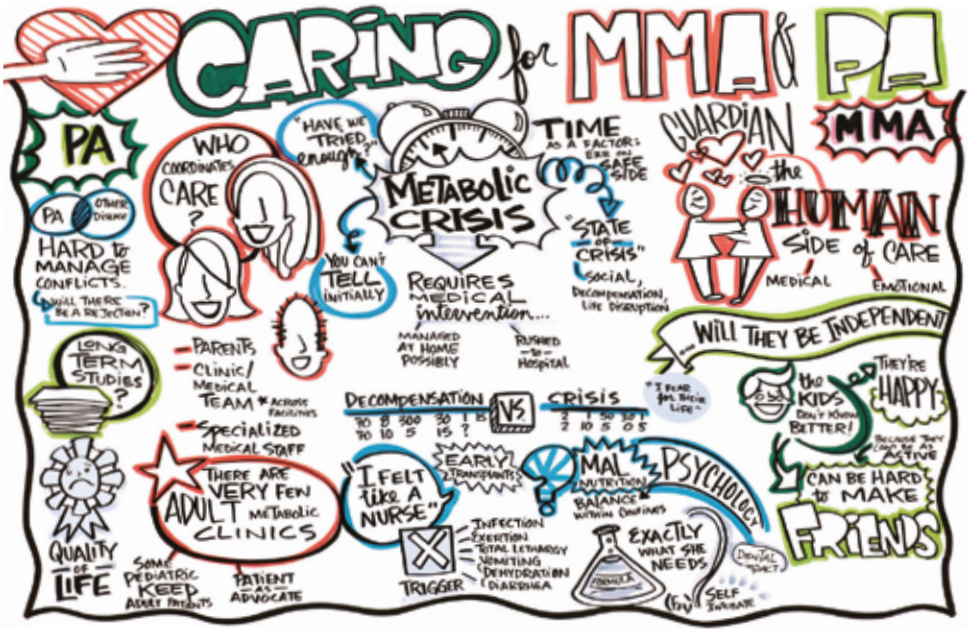
Chiesi
global rare diseases

Families Need New Hope



This graphic is from a workshop convened by HemoShear of parents sharing their children's experience with MMA and PA.

HemoShear is dedicated to improving the lives of patients with rare diseases.

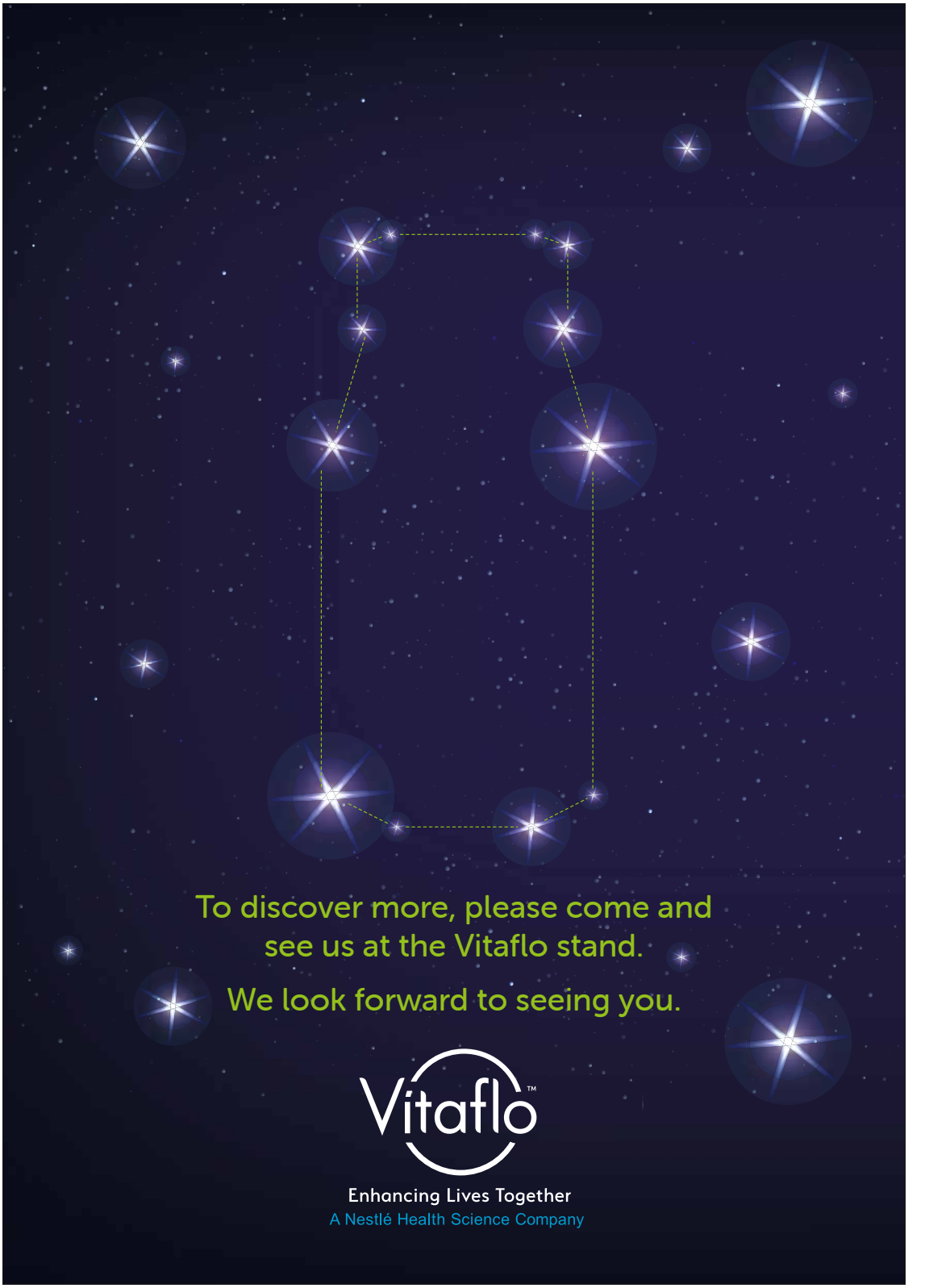
We are developing small molecule therapy HST5040 for the potential treatment of methylmalonic (MMA) and propionic acidemia (PA).

HELP REDUCE ORGANIC ACIDS
HERO STUDY

The **HERO** phase 2 clinical study of HST5040 is recruiting patients at select children's hospitals in the United States. More information can be found at clinicaltrials.gov (NCT04732429).

To learn more about HemoShear and be contacted about future studies, complete the form or reach out to Pat.Horn@Hemoshear.com.

HEMOSHEAR THERAPEUTICS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We look forward to seeing you.
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INNOVATION FOR THE BETTER

We are an Italian pharmaceutical company committed to providing doctors and patients with products that derive from incremental innovation and technological formulation, independently of whether they are drugs, food supplements or food for special medical purposes.

We stand out as reference point for medical specialists and patients by developing and offering state-of-the-art solutions that help improve therapeutic compliance, especially in the rare and niche disease area, while still ensuring that patients have full access to the pharmaceutical and medical food options they need.

We work alongside the medical and scientific community and together with international partners who hold high technological expertise to identify and deliver the best therapeutic solutions more quickly.

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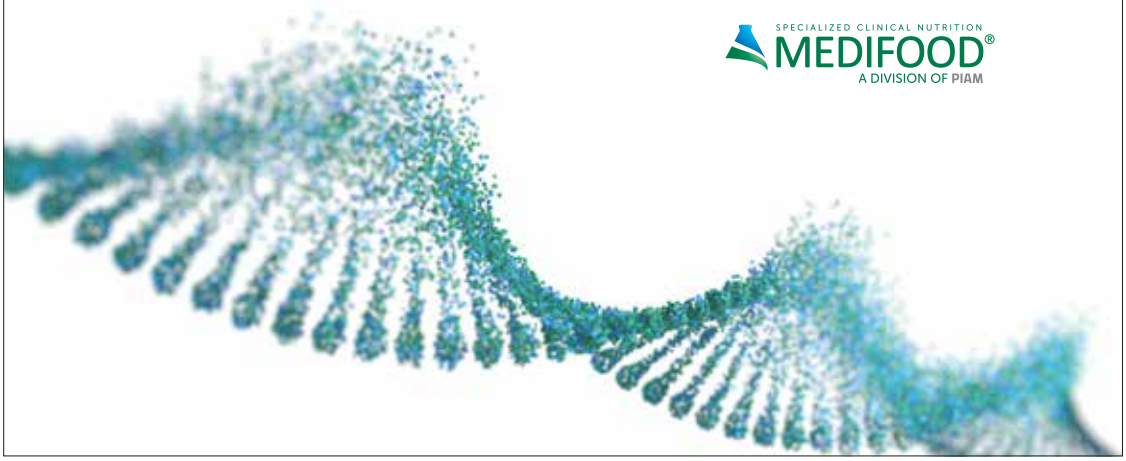

Mark Your Calendar!

SSIEM Annual Symposium 2023

29 August - 1 September 2023
Jerusalem, Israel

החברה הישראלית למחלות מטבוליות
Israel Society for Metabolic Diseases

Important Dates & Deadlines

Start Online Registration: 4th January 2023
Start Call for Abstracts: 16th January 2023
Abstract Submission Deadline: 19th April 2023

| NOTES |

NOTES

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We chase the *miracles of science*, with an enduring commitment to *better care for rare*.

We strive to:

- Break down the barriers to timely and accurate rare disease diagnoses
- Discover and develop new, innovative treatments that improve real-world outcomes
- Advocate for equitable access to medicines
- Elevate the voices of people living with rare diseases and support them across their lifelong journey

Come visit us at our booth

Sanofi-Aventis Deutschland GmbH - Lützowstraße 107
D-10785 Berlin – Germany
Approval Date: July 2022
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