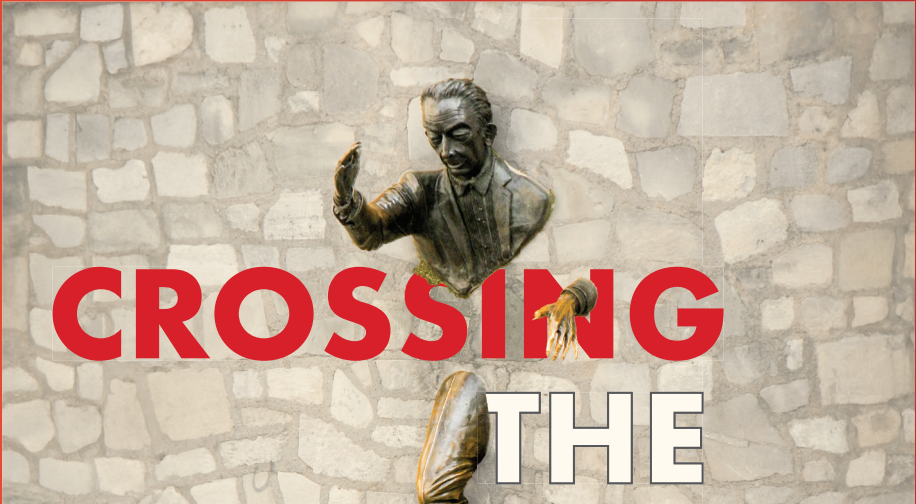


PROGRAMME

**APS**  
2024

# 37<sup>th</sup> Annual Conference and Seminar



# BARRIER

Inborn errors of metabolism  
affecting the brain

12.-15.03.2024

Kongress Palais Kassel

[www.events.aps-med.de](http://www.events.aps-med.de)

**Publisher**  
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# TABLE OF CONTENTS

APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR



Schedule .....	4
Welcome .....	5
Programm APS Stoffwechselfseminar .....	6
Dienstag, 12. März 2024 .....	6
Mittwoch, 13. März 2024 .....	6
Program APS Annual Conference .....	8
Wednesday, March 13, 2024 .....	8
Thursday, March 14, 2024 .....	10
Friday, March 15, 2024 .....	13
General information .....	15
Arrival in Kassel .....	16
Speakers information .....	17
Contact information .....	18
Sponsors of the APS Annual Conference .....	19
Site map .....	21

# SCHEDULE





## APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR

### TUESDAY, MARCH 12, 2024

17:45	Welcome + Evening Lecture	H4 Hotel, Gartensaal 1
19:30	Get Together / Dinner APS Seminar	H4 Hotel

### WEDNESDAY, MARCH 13, 2024

08:30-17:00	<b>APS Seminar</b>	Gesellschaftssaal
17:00-17:30	Satellite Symposium - 	Gesellschaftssaal
17:30-19:00	Satellite Symposium - <b>sanofi</b>	Bankettsaal
18:00-19:00	Satellite Symposium - 	Gesellschaftssaal
from 19:00	Get Together with Poster Viewing	Festsaal

### THURSDAY, MARCH 14, 2024

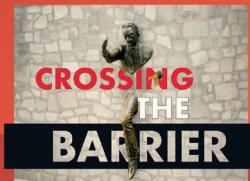
08:30-15:45	<b>APS Annual Conference</b>	Blauer Saal
15:45-17:00	Session of the Young Metabolic Society	Blauer Saal
17:00-18:30	APS general assembly	Blauer Saal
from 19:30	APS Dinner	Haupthalle, Kongress Palais

### FRIDAY, MARCH 15, 2024

08:30-13:30	<b>APS Annual Conference</b>	Blauer Saal
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# WELCOME

APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR



## Dear friends and colleagues,

It is our special pleasure to welcome you to the 37<sup>th</sup> APS conference in Kassel.

The field of inborn errors of metabolism is making huge progress in the development of new diagnostic procedures as well as therapeutic options. Neurometabolic disorders, affecting the brain, have in part been an exception to this, as the blood brain barrier, protecting the brain from harmful products, is a hurdle when it comes to treatment. The brain can be affected with many inborn errors of metabolism, either by toxins, energy deficiency or storage.

An excellent faculty of metabolic specialists and child neurologists will enhance our understanding of pathomechanisms, regional or age dependent brain vulnerability and secondary effects, which contribute to brain damage. We will hear upfront topics of clinical and diagnostic aspects as well as novel treatments including cargo enzymes, ASO and gene therapy for neurometabolic disorders.

The free oral communications and posters will be an asset to the main programme and we thank all authors for their active contributions. Please use the chances for personal exchanges and discussions and attend posters during the coffee breaks.

As a novelty along this conference, the *Young Metabolic Society* has organized one session within the main programme, providing updates on several selected neurometabolic disorders. We warmly welcome this initiative of the next generation of metabolic experts.

Our social event will be within the conference venue, and we hope to create an atmosphere that enhances professional discussions but also stimulates friendships and networking.

Best wishes,  
Prof. Dr. Barbara Plecko  
PD Dr. Anna Baghdasaryan, PhD

DIENSTAG, 12. MÄRZ 2024

H4 HOTEL, GARTENSAAL 1

- 17.45 Eröffnung und Vorsitz  
*Amelie Lotz-Havla (München)*
- 18.00 **Evening Lecture**  
Digitale Möglichkeiten in der Erkennung seltener Erkrankungen und in der Führung von Patienten mit chronischen Leiden  
*Michael Lang (Ulm)*
- 19.30 **Get Together / Dinner**

MITTWOCH, 13. MÄRZ 2024

GESELLSCHAFTSSAAL

Vorsitz: *Chris Mühlhausen (Göttingen)*

- 08.30 Basics der Liquordiagnostik  
*Thomas Opladen (Heidelberg)*
- 09.15 Mukopolysaccharidosen  
*Julia Hennermann (Mainz)*
- 10.00 **Kaffeepause**
- 10.30 Gruppenarbeit POL-Fall 1  
– *René Santer (Hamburg)*  
– *Chris Mühlhausen (Göttingen)*  
– *Sarah Grünert (Freiburg)*  
– *Amelie Lotz-Havla (München)*

# PROGRAMM

APS STOFFWECHSELSEMINAR



MITTWOCH, 13. MÄRZ 2024

GESELLSCHAFTSSAAL

**12.00 Mittagessen**

*Vorsitz: Sarah Grünert (Freiburg)*

**13.15** Pilotprojekte des Neugeborenen Screenings /  
Neugeborenen Screening der Zukunft  
*Esther Maier (München)*

**13.45** POL-Fall 2  
– *René Santer (Hamburg)*  
– *Chris Mühlhausen (Göttingen)*  
– *Sarah Grünert (Freiburg)*  
– *Amelie Lotz-Havla (München)*


**15.15 Kaffeepause**

*Vorsitz: René Santer (Hamburg)*

**15.45** Der interessante Stoffwechsel-Fall

**17.00** Verabschiedung  
*Amelie Lotz-Havla (München)*

WEDNESDAY, MARCH 13, 2024

17:00-17:30 **Satellite Symposium** with the friendly support of   
**Gesellschaftsaal**

Reformulating paediatric medicines to remove unwanted and harmful excipients  
*Stephen Tomlin (London, UK)*

17.30-19.00 **Satellite Symposium** with the friendly support of   
**Bankettsaal**

### Update Spingolipidosen

Spingolipidosen: What's good to know?  
*Ali Tunç Tuncel (Heidelberg, GER)*

Update vom WORLD-Symposium 2024  
*Mona Lindschau (Hamburg, GER)*

Follow-Up ASMD und Olipudase alfa  
*Eugen Mengel (Hochheim am Main, GER)*

18.00-19.00 **Satellite Symposium** with the friendly support of   
**Gesellschaftsaal**

**Hyperargininemia – An alien among urea cycle defects**  
*Chair: Julia Hennermann (Mainz, GER)*

Loargys – First approved therapy targeting hyperargininemia  
*Frank Rutsch (Münster, GER)*

How to use Loargys and first experience  
*Daniela Karall (Innsbruck, AT)*

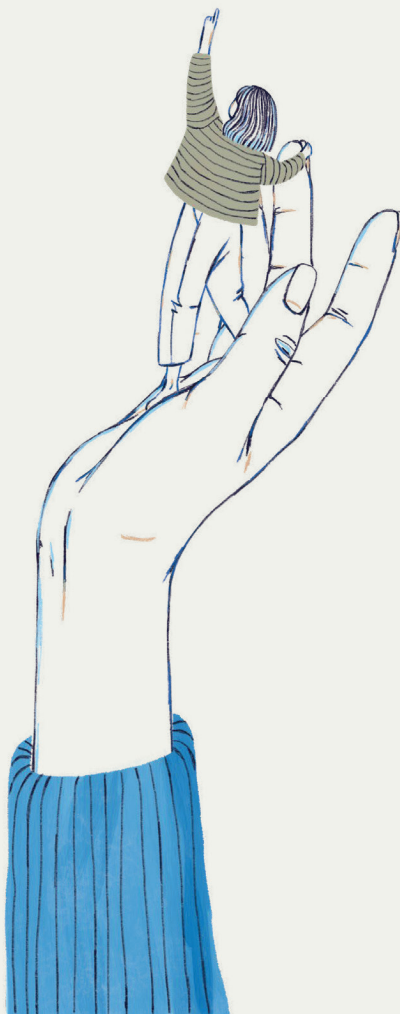
19.00 **Get together**  
Wine, finger food and poster viewing  
**Festsaal**



# ARGININ IM GRIFF PATIENTEN IM AUFWIND

**Loargys (Pegzilarginase)** ist die erste und einzige Behandlung, die modifizierend auf den Arginase-1-Mangel (ARG1-D) wirkt<sup>1</sup>.

- Loargys normalisiert bei Patienten mit ARG1-D rasch den Arginin-Plasmaspiegel und reduziert toxische Argininderivate<sup>2,3</sup>
- Loargys erzielt langanhaltende und klinisch bedeutsame Verbesserungen der funktionellen Ergebnisse bei einem überschaubaren Sicherheitsprofil<sup>2,3</sup>
- Loargys hat das Potenzial, die Behandlung von ARG1-D bei Patienten mit bislang wenigen klinischen Optionen zu revolutionieren<sup>1</sup>



1. Diaz GA, et al. The role and control of arginine levels in arginase 1 deficiency. *J Inher Metab Dis.* 2023 Jan;46(1):3-14. | 2. Sanchez Russo R et al. Pegzilarginase in Arginase 1 Deficiency: Results of the PEACE Pivotal Phase 3 Clinical Trial (PAS4A-2674). Oral presentation at the SSIEM Annual Symposium, Freiburg, Germany 30 August – 2 September 2022. | 3. Sanchez Russo R. Safety and Tolerability of Pegzilarginase for Arginase 1 Deficiency in the PEACE Pivotal Phase 2 Clinical Trial. Poster presented at the SSIEM Annual Symposium, Freiburg, Germany, 30 August – 2 September 2022.

**Loargys 5mg/ml Lösung zur Injektion/Infusion. Darreichungsform:** Jede 0,4ml Durchstechflasche enthält 2 mg Pegzilarginase. **Anwendungsgebiet:** Loargys ist indiziert für die Behandlung von Arginase-1-Mangel (ARG1-D), auch bekannt als Hyperargininämie, bei Erwachsenen, Jugendlichen und Kindern ab 2 Jahren. **Dosierung und Verabreichung:** Loargys sollte durch intravenöse Infusion oder subkutane Injektion gegeben werden, wobei die Dosis gleich ist. Gegebenenfalls kann nach mindestens 8-wöchiger Behandlung eine subkutane Anwendung durch den Patienten oder die Betreuungsperson in Erwägung gezogen werden, sobald eine stabile Erhaltungsdosis bestimmt wurde und das Risiko für Überempfindlichkeitsreaktionen als gering eingestuft wird. Die empfohlene Anfangsdosis von Loargys beträgt 0,1mg/kg pro Woche. Die Dosis kann in Schritten von 0,05mg/kg erhöht oder verringert werden, um die therapeutischen Ziele zu erreichen. **Gegenanzeigen:** Überempfindlichkeit gegen den Wirkstoff oder einen der sonstigen Bestandteile. **Warnhinweise und Vorsichtsmaßnahmen:** Bei mit Loargys behandelten Patienten traten Überempfindlichkeitsreaktionen auf. Die Reaktionen traten im Allgemeinen bei den ersten Dosen auf. **Fertilität, Schwangerschaft und Stillzeit:** Die Anwendung von Pegzilarginase während der Schwangerschaft und bei gebärfähigen Frauen, die keine Empfängnisverhütung anwenden, wird nicht empfohlen. **Nebenwirkungen:** Die am häufigsten berichteten Nebenwirkungen bei Patienten in den klinischen Studien waren Überempfindlichkeit und Reaktionen an der Injektionsstelle. Verschreibungspflichtig. Pharmazeutischer Unternehmer: Immedica Pharma AB, Solnavägen 3H, 113 63 Stockholm, Schweden. Weitere Informationen siehe Fach- und Gebrauchsinformation von Loargys. **Stand:** Dezember 2023.

▼ Dieses Arzneimittel unterliegt einer zusätzlichen Überwachung.

THURSDAY, MARCH 14, 2024

**BLAUER SAAL**

08.30-08.40 **Welcome and conference outline**

*Barbara Plecko (Graz)*

08.40-10.00 **What makes the human brain a special organ?**

Chair: Barbara Plecko (Graz, AT),  
Johannes Häberle (Zurich, CH)

08.40-9.05 Cellular and molecular mechanisms of the blood brain barrier

*Christine Stadelmann-Nessler (Göttingen, GER)*

09.05-9.35 Inborn errors of metabolism, neurotoxicity and age-dependent brain vulnerability

*Ángeles García Cazorla (Barcelona, ES)*

9.35-10.00 The complex pathophysiology of lysosomal storage disorders

*Frances Platt (Oxford, UK)*

10.00-10.30 **Coffee Break | Posters | Industrial Exhibition**

10.30-12.30 **Odysseus´ changed decisions  
– quicker ways to diagnose IEM**

Chair: Gwendolyn Gramer (Hamburg, GER),  
Peter Freisinger (Reutlingen, GER)

10.30-11.00 Efficacy of newborn screening for the prevention of disease manifestation in neurometabolic disorders

*Ulrike Mütze (Heidelberg, GER)*

11.00-11.30 Neurometabolic Disorders – a signs and symptoms approach

*Lucia Abela (Zurich, CH)*

# PROGRAMME

APS 37<sup>TH</sup> ANNUAL CONFERENCE



THURSDAY, MARCH 14, 2024

BLAUER SAAL

- 11.30-12.00 Do we still need biomarkers in the diagnostic work-up of children with unclear neurologic disease?  
*Saskia Wortmann (Salzburg, AT)*
- 12.00-12.30 Dominant or recessive? Functional variant interpretation in genetic diagnostics  
*Johannes Zschocke (Innsbruck, AT)*
- 12.30-13.30 **Lunchbreak and Poster Viewing**
- 13.30-14.30 **Update on Leukodystrophies**  
*Chair: Steffie Dreha-Kulaczewski, Göttingen (GER), Felix Distelmaier, Düsseldorf (GER)*
- 13.30-14.00 Metabolic and inflammatory aspects in Leukodystrophies  
*Jutta Gärtner (Göttingen, GER)*
- 14.00-14.30 The change in the diagnostic and therapeutic paradigm with a focus on X-ALD and MLD  
*Nicole Wolf (Amsterdam, NL)*
- 14.30-15.20 **Free communications I**  
*Chair: Anibh Das (Hannover, GER), Julien Park (Münster, GER)*
- FC 01-01 Improving the diagnostic performance of testing for Metachromatic Leukodystrophy: a new LC-MS/MS method including normalization of urinary sulfatide concentration to sphingomyelin  
*Manuela Peschka (Hamburg, GER)*
- FC 01-02 First newborns to receive gene therapy after prospective newborn screening for metachromatic leukodystrophy  
*Lucia Laugwitz (Tübingen, GER)*

THURSDAY, MARCH 14, 2024

**BLAUER SAAL**

FC 01-03 The clinical, neuroimaging and metabolic footprint of the neurodevelopmental disorder caused by monoallelic HK1 variants – a differential diagnosis for GLUT1 deficiency syndrome and mitochondrial disorders  
*Saskia Wortmann (Salzburg, AT)*

FC 01-04 Severity prediction model for nonketotic hyperglycinemia: validation approach  
*Oya Kuseyri Hübschmann (Heidelberg, GER)*

15.20-15.45 **Coffeebreak and Poster Viewing**

15.45 -17.00 **Session of the Young Metabolic Society**  
*Chair: Julia Neugebauer (Berlin, GER),  
Ali Tunç Tuncel (Heidelberg, GER)*

GA I-Update  
*Eva Marie Charlotte Märtner (Munich, GER)*

Enzyme Replacement Therapy in PKU  
*Aloisa Stadlhofer (Hamburg, GER)*

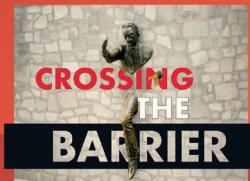
Metabolic Epilepsies  
*Lara Marten (Göttingen, GER)*

17.00-18.30 **APS general assembly**

19.30 **APS Dinner**  
**Haupthalle, Kongress Palais**

# PROGRAMME

APS 37<sup>TH</sup> ANNUAL CONFERENCE



FRIDAY, MARCH 15, 2024

BLAUER SAAL

**8.30-10.00 Free communications II**

*Chair: Anna Baghdasaryan (Graz, AT),  
Matthias Gautschi (Bern, CH)*

**FC 02-01** Impact of liver transplantation on health outcomes in urea cycle disorders

*Roland Posset (Heidelberg, GER)*

**FC 02-02** Clinical and genomic testing outcomes in children with severe or recurrent hypoglycaemia at The Royal Children's Hospital, Melbourne, 2016-2021

*Oliver Heath (Salzburg, AT)*

**FC 02-03** Genetic landscape of paediatric acute liver failure of indeterminate origin

*Alyssa Bianzano (Heidelberg, GER)*

**FC 02-04** Repurposing SGLT2 inhibitors: treatment of renal proximal tubulopathy in Fanconi-Bickel syndrome with empagliflozin

*Ruben J. Overduin (Groningen, NL)*

**FC 02-05** Added diagnostic value of (long-read) genome sequencing and clinical implementation within the Modellvoraben §64e

*Tobias Haack (Tübingen, GER)*

**FC 02-06** Free carnitine concentrations and biochemical parameters in medium-chain acyl-CoA dehydrogenase deficiency:

Genotype-phenotype correlation

*Katharina J. Weiß (München, GER)*

**FC 02-07** Niemann-Pick type B disease (ASMD): a series of four cases and first results from enzyme replacement therapy (ERT) with olipudase alfa

*Stephan vom Dahl (Düsseldorf, GER)*

**FC 02-08** ASS1 deficiency is associated with impaired neuronal differentiation in zebrafish larvae

*Marie J. Seidl (Heidelberg, GER)*

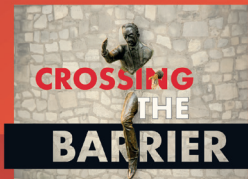
FRIDAY, MARCH 15, 2024

**BLAUER SAAL**

- 10.00-10.30 **Coffeebreak and Poster Viewing**
- 10.30-12.30 **Therapeutic Innovation Pipeline in Neurometabolic Disorders**  
*Chair: Ute Spiekerkötter, Freiburg (GER),  
Thomas Opladen, Heidelberg (GER)*
- 10.30-11.00 Drug repurposing in mitochondrial disorders - and beyond  
*Markus Schülke (Berlin, GER)*
- 11.00-11.30 The blood-brain barrier and enzyme replacement therapies  
– devices versus cargo  
*Angela Schulz (Hamburg)  
Julia Hennermann (Mainz, GER)*
- 11.30-12.00 Approaches to studying innovative therapies for ultrarare  
diseases  
*Alfonso de Oyarzábal Sanz (Barcelona, ES)*
- 12.00-12.30 Genetic therapies for neurometabolic disorders  
*Marlen Lauffer (Leiden, NL)*
- 12.30-12.40 **Prizes and Farewell**
- 12.40-13.30 **Lunch and Poster Viewing**

# GENERAL INFORMATION

APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR



## CONFERENCE VENUE

Kongress Palais Kassel  
Holger-Börner-Platz 1  
34119 Kassel  
Germany

Phone: +49 561 707702  
Email: [info@kassel-marketing.de](mailto:info@kassel-marketing.de)  
[www.kongress-palais.de](http://www.kongress-palais.de)

## ARRIVAL BY CAR

The Kassel Kongress Palais is located in the middle of Germany and can be reached from all directions in Europe by the shortest route. Parking is available in the Parkhaus Kongress Palais / Kattenstraße with 109 spaces. Further public parking spaces are available in the immediate vicinity.

## ARRIVAL BY TRAIN

ICE Hamburg / Bremen – Kassel – Frankfurt – Stuttgart or Basel / Zurich,  
ICE Kiel – Hamburg – Kassel – Würzburg – Munich or Nuremberg / Vienna,  
ICE Berlin – Braunschweig – Kassel – Frankfurt – Munich,  
IC Hamburg – Kassel – Frankfurt – Konstanz, IC Düsseldorf – Kassel – Erfurt – Weimar

## ARRIVAL BY PLANE

Frankfurt Airport provides a direct connection to the European air network. German airports are served particularly frequently, and the flight time within Germany is less than one hour. From Frankfurt Airport, you can get directly to Kassel by train in about two hours.

## CONFERENCE FEES

<b>Registration for the APS Annual Conference</b>	<b>by February 15<sup>th</sup></b>	<b>from February 16<sup>th</sup></b>
APS Members	€ 65,-	€ 90,-
Non-Members	€ 100,-	€ 125,-
APS Dinner	€ 20,-	€ 20,-

Students and PhD candidates do not have to pay this fee on presentation of a corresponding certificate when registering.

Please register online at [www.events.aps-med.de](http://www.events.aps-med.de).

Participation in the „APS Stoffwechsel Seminar“ is free of charge for physicians, dieticians and metabolic lab personell.

# ARRIVAL IN KASSEL



**Tram 4** in the direction of Kaufungen-Papierfabrik  
4 min, 3 stops to Kassel Kongress Palais/Stadthalle  
Alternatives every 15 minutes



# SPEAKERS INFORMATION

APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR



## **Conflict of interest**

We would like to point out that the speakers are required to present in a product and service-neutral manner. Possible conflicts of interest must be announced at the beginning of the presentation.



## **Conference language**

Official language of the APS Annual Meeting is English, the APS Seminar will be held in German.



## **Compatibility**

Please submit your presentation as a PC-compatible file on a data storage device (memory stick). After saving it on the data storage device, please make sure the presentation is running smoothly. Speakers are responsible for their presentation's compatibility.



## **Media center**

Please hand in your data storage device at the "media center" at the registration desk 2 hours before your presentation.



## **Submission via e-mail**

You can also send your presentation in advance as an e-mail attachment to [aps@studio12.co.at](mailto:aps@studio12.co.at) (by March 6, 2024 at the latest).



## **MacOS**

Mac users are urgently requested to save their presentation on the data storage device in a PC-compatible way and ensure that it will run without issue.

### HOST

#### APS Annual Conference

##### **Univ.-Prof. Dr. Barbara Plecko**

Univ. Klinik für Kinder- und Jugendheilkunde  
Auenbruggerplatz 34/2, 8036 Graz, Austria  
E-Mail: barbara.plecko@medunigraz.at

##### **Univ. FÄ Priv.-Doz. Dr. Anna Baghdasaryan, PhD**

Univ. Klinik für Kinder- und Jugendheilkunde  
Auenbruggerplatz 34/2, 8036 Graz, Austria  
E-Mail: a.baghdasaryan@medunigraz.at

#### APS Seminar

##### **Dr. med. Amelie Lotz-Havla**

LMU Klinikum, Kinderklinik und Kinderpoliklinik  
im Dr. von Haunerschen Kinderspital  
Lindwurmstraße 4 | 80337 München  
E-Mail: Amelie.Lotz@med.uni-muenchen.de

##### **Prof. Dr. Sarah Grünert**

Klinik für Allgemeine Kinder- und Jugendmedizin  
Universitätsklinikum Freiburg  
Heiliggeiststraße 1, 79106 Freiburg  
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#### Congress Organiser

##### **studio12 gmbh**

Kaiser-Josef-Straße 9  
6020 Innsbruck, Austria  
Phone: +43-(0)512-890438  
E-Mail: aps@studio12.co.at

#### Education points

The recognition of the Annual Conference and Seminar as a certified event for Continuing Medical Education has been approved by the Landesärztekammer Hessen. The credit points are allocated as follows:

##### **APS Stoffwechselfseminar**

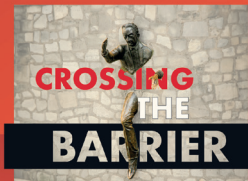
12.03.2024: 2 Points      13.03.2024: 7 Points

##### **APS Annual Conference**

14.03.2024: 7 Points      15.03.2024: 4 Points

# SPONSORS

## APS 37<sup>TH</sup> ANNUAL CONFERENCE AND SEMINAR



Die Inhalte dieser Veranstaltung werden produkt- und dienstleistungsneutral gestaltet. Wir bestätigen, dass die wissenschaftliche Leitung und die Referenten potentielle Interessenkonflikte gegenüber den Teilnehmern offenlegen. Folgende Firmen treten als Sponsoren auf:

POSITION	FIRMA	FÖRDERSUMME (€)
Ausstellungsstand	Advanz Pharma Germany GmbH	5.500,00 €
Ausstellungsstand	Ajinomotto Cambrooke	5.500,00 €
Ausstellungsstand	Amicus Therapeutics	5.500,00 €
Stornogebühren	APR Deutschland GmbH	2.750,00 €
Ausstellungsstand	BioMarin Deutschland GmbH	11.000,00 €
Symposium	CENOTE Pharma GmbH	3.000,00 €
Ausstellungsstand	Chiesi Pharmaceuticals GmbH	5.500,00 €
Ausstellungsstand	Dipharma Arzneimittel GmbH	6.500,00 €
Ausstellungsstand + Symposium + Inserat	Immedica Pharma	17.500,00 €
Ausstellungsstand	Kanso	5.500,00 €
Ausstellungsstand	Laborservice Onken GmbH	5.500,00 €
Ausstellungsstand	Lucane Pharma (MAH Eurocept International B.V.) + Apocare	5.500,00 €
Ausstellungsstand	metaX Institut für Diätetik GmbH	5.500,00 €
Ausstellungsstand	Nutricia Metabolics	16.500,00 €
Ausstellungsstand	POA Pharma GmbH	5.500,00 €
Ausstellungsstand	PTC Therapeutics	5.500,00 €
Ausstellungsstand + Symposium	Sanofi-Aventis Deutschland GmbH	10.500,00 €
Ausstellungsstand	Sobi	5.500,00 €
Ausstellungsstand	Takeda Pharma Vertrieb GmbH & Co. KG	5.500,00 €
Ausstellungsstand	Ultragenyx Germany GmbH	11.000,00 €
Ausstellungsstand	Vitaflo Deutschland GmbH	5.500,00 €
	<b>Summe</b>	<b>150.250,00 €</b>

**APS-SEMINAR:** Für das Seminar findet kein Sponsoring statt. Die Veranstaltung wird ausschließlich durch die APS selbst finanziert. Höhe der Gesamtaufwendungen: 8.000 EUR.

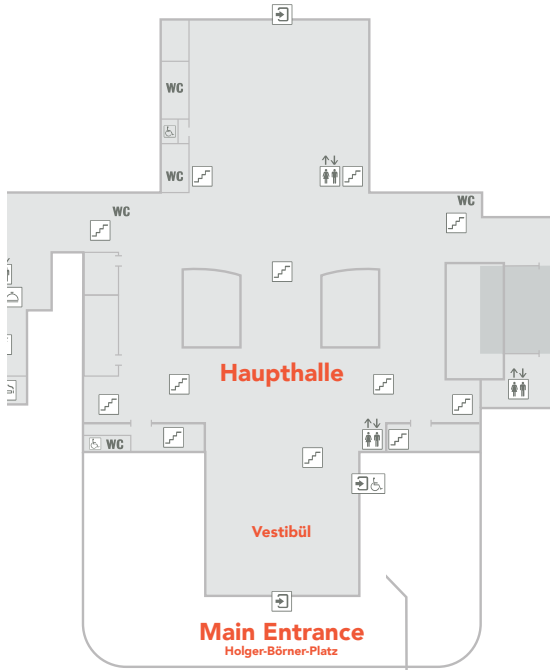
(Disclosure of sponsorship services in accordance with the FSA Code as part of the expanded transparency requirement for the support of congress events)

# SPONSORS

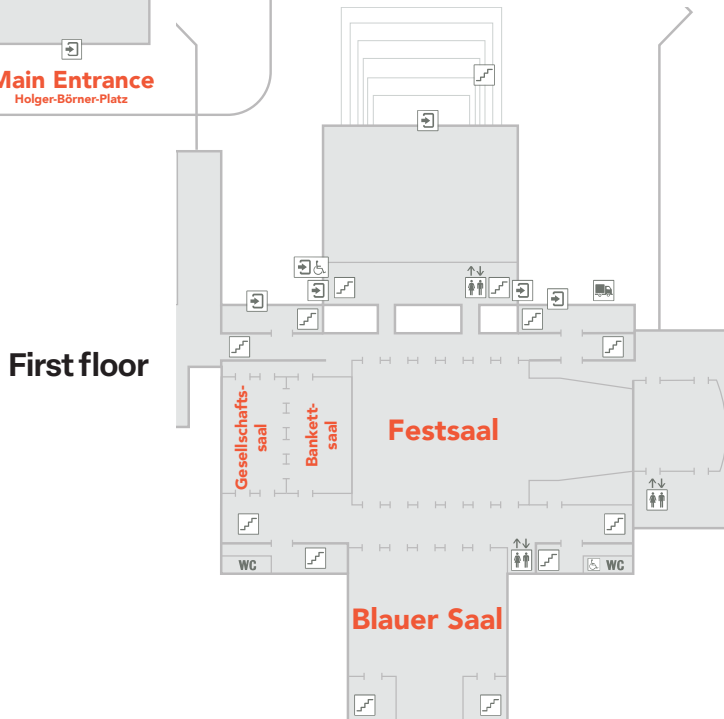
APS 37<sup>TH</sup> ANNUAL CONFERENCE



# SITE MAP



Ground floor



First floor







# 37<sup>th</sup> Annual Conference and Seminar

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