



## MINUTES

### UEMS MJC RUD Annual Meeting

<b>Date and time:</b> November 15, 2022 17:00-19:00				<b>Protocol No 3/2022</b>
<b>Place:</b> online				
<b>Attendees:</b> Bela Melegh Artur Beke Serdar Ceylaner Alessandra Renieri Ambrogio Fassina Joel Ferri Pat Twomey Domenica Taruscio John Firth Olle Hollertz George Petrikkos Eva Tienssu Janson Daniel Hohl Marek Bolanowski Jean Calleja Agius Anna Nikulina Oleg Kvlividze Josanne Vassallo Leah Matthews		<b>Excused:</b> Maeve Durkan Marek Bolanowski Rijk OB Gans Risto Lapatto		
	<b>Point to discuss</b>	<b>Discussion</b>	<b>Decisions</b>	<b>Reference person</b>
<b>1.</b>	Opening of the meeting by BM and approval of agenda	Meeting is opened at 17:00 by BM. PT requested to postpone point 9., because it was discussed shortly before the MJC RUD meeting at the Section of Laboratory Medicine.	Agenda approved.  BM agreed and asked that the supporting documents like Explanatory Notes from the Section of Laboratory Medicine be provided later on. *	BM

2.	Approval of the Minutes of the 2021 Annual Meeting, Limassol	BM outlined the minutes from the 2021 Annual Meeting in Limassol.	Minutes approved	BM
3.	Annual report of the President (year 2021)	BM outlined the Annual Report of the President.	Annual Report approved	BM
4.	Annual report of the Treasurer (year 2021)	AB presented slides on the Annual Report.	Annual Report approved	AB
5.	EU Exam “Fundamentals of Rare and Undiagnosed Diseases” issues	To proceed with this task, the following steps are proposed: - Description of the exam (volunteers are needed to constitute a 10-20 pages long text) - establishment of Enlarged Exam Steering Committee with the complete and exclusive authorization of the coordination and management of all exam tasks, with a 4 year long mandate (starting from January 2023). - organization of a remote (online) trial exam in early 2023	Approved  Approved  Approved	BM
6.	European Neuroendocrine Tumor Society (ENETS)	Presentation by ENETS	BM congratulated on behalf of MCJ RUD and offered his further support.	ETJ, LM
7.	WG for Rare Diseases of Bone Tissues of the Head (WGRDBTH)	BM presented the letter of OMFS section.	After extensive discussion the proposal of OMFS was unanimously approved and JF was elected as head of WG of RDBTH.	BM
8.	European Board of Rare and Undiagnosed Diseases (EBRUD) issues	The delegates already received the letter circulated amongst the UEMS bodies calling for nominations to the EBRUD.	After nominations the MJC RUD will continue with EBRUD preparation works according to the already approved organizational chart.	BM

<b>9.</b>	WG for Laboratory Genetics, ETR for Clinical Laboratory Genetics	Postponed by the request from PT.	Request approved. Waiting for explanatory notes. *	BM
<b>10.</b>	CME/CPD	Preliminary draft by BM.	Further discussion is needed.	BM
<b>11.</b>	AOB	-		
<b>12.</b>	Closing of meeting	Meeting is closed at 19:00		

\* Letter from Section of Laboratory Medicine by Nov. 18, 2022 received electronically on the same day (attached).

Minutes taken by BM

## **AGENDA**

with supplementary notes

### **Annual Meeting of MJC RUD**

15 November, 2022

online meeting, 17.00 – 19.00 hrs

#### **1., Opening, approval of the Agenda**

#### **2., Approval of the Minutes of the 2021 Annual Meeting, Limassol**

#### **3., Annual report of the President (year 2021)**

#### **4., Annual report of the Treasurer (year 2021)**

#### **5., EU Exam “Fundamentals of Rare and Undiagnosed Diseases” issues**

To proceed with this task, the following steps are proposed:

- Description of the exam (volunteers are needed to constitute a 10-20 pages long text)
- establishment of Enlarged Exam Steering Committee with the complete and exclusive authorization of the coordination and management of all exam tasks, with a 4 year long mandate (starting from January 2023).
- organization of a remote (online) trial exam in early 2023

#### **6., European Neuroendocrine Tumor Society (ENETS)**

- a representative is invited to present the current status

#### **7., WG for Rare Diseases of Bone Tissues of the Head (WGRDBTH)**

- letter of OMFS Section; due to personal changes we have to reshuffle the current WG RDBTH according to the proposal of the Bureau of the OMFS Section

#### **8., European Board of Rare and Undiagnosed Diseases (EBRUD) issues**

- preparatory works are on the way, letter to the UEMS Bodies

#### **9., WG for Laboratory Genetics, ETR for Clinical Laboratory Genetics**

- proposal for the creation of this new WG, with the long-term goal of the construction of an ETR

#### **10., CME/CPD**

- new approach is proposed: development of a system for EU Exam holders

#### **11., AOB**

MINUTES  
 UEMS MJC of Rare and Undiagnosed Diseases (RUD)  
 2020-2021 Annual Meeting Limassol

<b>Date:</b> October 22, 2021				<b>Protocol No 1/2021</b>
<b>Place:</b> Limassol / teleconference				
<b>Attendees:</b> Bela Melegh, President Artur Beke Alessandra Renieri, Secretary Domenica Taruscio Joel Ferri Serdar Ceylander Marko Stojanovic Oleg Kvlividze Risto Lapatto Umut Akyol Josanne Vassallo Malik Maharramli Vesna Kusec Patrick Twomey Ilia Nadareishvili Ipad de Daniel?				
	<b>Point to discuss</b>	<b>Discussion</b>	<b>Decisions</b>	<b>Reference person</b>
<b>1</b>	Opening of the meeting by Bela Melegh and Approval of agenda	Meeting is open at 9:00.	Agenda approved (Supplement 1)	Bela
<b>2</b>	"2018 Minutes", and "2020 Activity report"	Bela presented "2018 Minutes" (Supplement 2.), and "2020 Activity report" (Attachment 1)	Unanimously approved	Bela
<b>3</b>	The 2019-2020 annual report (President)	Bela presented the Annual Report (Supplement 3 and 4). There is no MJC RUD web at the moment. An MJC RUD membership registry revision is on the way (with Marine Boulard).	Reports unanimously approved. Funds will be requested from UEMS Exec for web and EBRUD launch. We will contact sections which are not represented in the MJC RUD, and ask for delegates.	Bela
<b>4</b>	Annual report of Treasurer	Artur presented the finances annual report (Supplement 5)	Unanimously approved	Artur

<b>5</b>	Undiagnosed Disease Network International (UDNI)	Domenica Taruscio presented the Undiagnosed Disease Network International (UDNI) as Strategic partner of MJC RUD. (Attachment 2) Alessandra proposed that every member as MJC RUD become also member of UDNI	There are already MJC RUD members in UDNI. All the other members are invited to submit the application to UDNI	Domenica
<b>6</b>	Rare diseases of calcified tissues of the head and neck	Joel Ferri presented talk with cases of rare diseases of calcified tissues of the head and neck. Bela proposed to Joel to build a dedicated Working Group (Attachment 3)	Creation of working group unanimously approved. Joel Ferri is the head of this WG	Joel
<b>7</b>	European Board of Rare and Undiagnosed Diseases (EBRUD)	Bela presented a draft (Supplement 6). He already contacted several external partners including Matt Bolz-Johnson and Sharon Ashton from the EURORDIS	The goal is to prepare the EBRUD materials to the 2022 Council. Funds will be requested for creation of EBRUD and exam.	Bela
<b>8</b>	European Exam in Rare and Undiagnosed Diseases	An exam committee should be created, as part of the MJC RUD and/or EBRUD.	Launch of European Exam preferably in 2022.	Bela
<b>9</b>	AOB	We will proceed with the MJC RUD membership registry housekeeping.	Bela and his secretary will work on this topic with Marine Boulard.	-
<b>10</b>	Closing meeting	Meeting is closed at 11:00		Alessandra

## **Annual Report of the President – 2021**

10 October, 2022

The calendar year of 2021 was the 5th full year in the history of the MJC RUD. The continuing Covid 19 pandemic strongly influenced our activity, however, the activity improved and we had a productive year. In short, the calendar year of 2021 was about the organizations and preparatory works.

### **Membership**

According to the last control, we had a total of 47 members from 27 Sections (OMFS, IM, Ophthalmology, Paediatrics, Psychiatry, Rheumatology, Medical Genetics, Neurology, Paediatric Surgery, Pharmacology, and Rehabilitation). They together were from 21 countries, and 3 additional were non EU Nations (Armenia, Georgia, Turkey). The following NMAs have delegates: Austria, Georgia, Greece, Spain, The Netherlands, Turkey. However, some members indicated their resignation, indirect evidence suggest that some of the members on the list are not active. The process of “housekeeping” is still on the way with the assistance of the UEMS Coordination.

### **The Bureau**

Electronic votes were organized by the Coordination in 2019-20; taken together, Dr. Domenica Taruscio was elected as vice-president, and Dr. Artur Beke was elected as treasurer, the president was re-elected. All 4 yrs office terms were agreed to start in 2021 January lasting till December of 2024. Thereby the Bureau consists of the president, secretary, treasurer, and 2 vice presidents with office terms till December of 2024.

### **MJC RUD Meetings**

There was an onsite/e-remote official membership meeting in association of the Limassol Council Meeting (22 October, 2021), which was a combined meeting for years 2020-2021. Besides this, well balanced activity was conducted also via emails.

#### **Main focus in 2020 and after**

As the Council approved the ETR of Rare and Undiagnosed Diseases (79.95 % support), Rare Adult Solid Cancer (68.42% support), and the ENETS ETR (88.89% support) in 2019, we focused on organization of future exam, and focused on the possible extension of training activities. Thus, the priorities included the preparatory phase of the European Board of Rare and Undiagnosed Diseases (EBRUD) and then the European Exam in fundamentals of rare diseases.

Working Group for Rare Diseases of Calcified Tissues of Head (WG RDCTH) was established with coordination of Joel Ferri from OMFS Section.

Besides this, intensive partnering was initiated with the EURORDIS.

### **Home page**

Historically, our the web was associated to the Section of Medical Genetics (SMG) as a courtesy of the provider to Dr Melegh, SMG president. This was not available after 2021 January, as the term of Dr Melegh expired and the new SMG Bureau suspended this

courtesy service. Thanks to the help of the UMES Exec, we reorganized the website in 2021, and we have a new profile (<https://mjcrud-uems.pt.hu>).

### **Finances**

Thanks to the aid of the UEMS Exec, we received a 7,000 Euros refundable loan from the UEMS budget. This sum, and the income from the event reviews, resulted in a closing balance of 7,576.97 Euros in 31 December, 2021, we already paid 300 Euros CESMA membership contribution for 2022. The president evaluated 7 life events for the CESMA.

### **Participation in other activities**

Lecture was presented at the 10th meeting of Undiagnosed Network International (UDNI) summarizing our main goals. The president participated in the work of the CESMA, and continued with the EACCME "Training the reviewers" working group.



Dr. Béla Melegh  
professor  
MJC RUD President



## **ANNUAL REPORT OF TREASURER**

UEMS MULTIDISCIPLINARY JOINT COMMITTEE ON RARE AND UNDIAGNOSED DISEASES

FINANCIAL STATEMENT SUMMARY FOR CALENDAR YEAR 2021

**Opening balance as of January 1<sup>st</sup> 2021** **0.00 EUR**

### **Income:**

EACCME REIMBT FEES REVIEWS 1ST SEMESTER 200.00 EUR  
/European Accreditation Council for Continuing Medical Education/  
2019 DATE VALEUR : 13/04/2021

EACCME REIMBT FEES REVIEWS 2ST SEMESTER 1,100.00 EUR  
/European Accreditation Council for Continuing Medical Education/  
2019 DATE VALEUR : 13/04/2021

EACCME REIMBT FEES REVIEWS FEES 2ND SEM 20 200.00 EUR  
European Accreditation Council for Continuing Medical Education/  
DATE VALEUR : 16/11/2021

EACCME REIMBT FEES REVIEWS FEES 1ST SEM 20 450.00 EUR  
European Accreditation Council for Continuing Medical Education/  
DATE VALEUR : 16/11/2021

UEMS LOAN 2021 7,000.00 EUR  
DATE VALEUR : 30/11/2021

**Total income** **8,950.00 EUR**

### **Expenditure:**

**UEMS CESMA contribution** - 300.00 EUR  
/Council for European Specialists Medical Assessment/

**UEMS Council Meeting - Limassol - 2021.10.22** - 962,13 EUR  
(Reimbursement Bela Melegh)

**Bank charges**

REDEVANCE TRIMESTRIELLE - 20.00 EUR

REDEVANCE TRIMESTRIELLE - 30.00 EUR

REDEVANCE TRIMESTRIELLE - 30.00 EUR

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REDEVANCE TRIMESTRIELLE - 30.00 EUR

INTERETS NETS : DETAILS VOIR ANN - 0.28 EUR

INTERETS NETS : DETAILS VOIR ANN - 0.32 EUR

INTERETS NETS : DETAILS VOIR ANN - 0.30 EUR

**Total expenditure** - **1,373.03 EUR**

**Closing balance as of December 31<sup>st</sup> 2021** **7,576.97 EUR**

Prepared by Artur Beke MD PhD

Treasurer, Multidisciplinary Joint Committee on Rare and Undiagnosed Diseases

31.12.2021

Letter from OMFS Bureau:

To

Dr. Melegh Béla

melegh.bela@pte.hu

MJC Rare and Undiagnosed Diseases (RUD)

October 24, 2022

Dear Colleague,

I am writing to you regarding the benefit to UEMS and to European patients of our friend and colleague Joel Ferri being able to continue his excellent work as the OMFS Section representative on the MJC RUD.

Dr Ferri is no longer a delegate to the OMFS Section, but we would be grateful if you could use your ability within the UEMS Rules of Procedure to invite him to be an observer on the MJC RUD.

If you confirm that you are happy to co-opt Dr Ferri onto the MJC RUD, we will invite him to attend the OMFS Section as a representative of the MJC RUD and an observer to the OMFS Section.

Patrick Magennis  
President OMFS Section

Hans-Peter Ulrich  
Secretary/Treasurer OMFS Section

To Presidents & Secretaries UEMS Specialist Sections, European Boards, MJC, Divisions

Dear Colleagues,

15 November, 2022

As you might be aware, at the UEMS Council of 29-30. April of 2022, creation of the European Board of Rare and Undiagnosed Diseases (EBRUD) was approved and the MJC RUD was thus authorized to proceed with the organization of the EBRUD, according to the RoP of the UEMS.

The MJC RUD team consulted the UEMS Enlarged Executive and already initiated the preparatory works.

It is an essential element in our Statutes and RoP, that when a European Board creation is approved by the UEMS Council, the MJC has to inform its MJC members (Sections and other bodies) and each interested UEMS body (Section, MJC) may nominate a delegate to the European Board. This nomination is not restricted to the Section's delegate to the MJC. Furthermore, with regard to the composition of the European Board: „Each delegation to a Board shall consist in 2 members, one from the Section and one from the recognized academic or Scientific Society of the country“, provided that the nomination conditions apply to each member.

During the preparatory works that have been performed so far, we have received nominations from Sections, but none from scientific societies yet. We would highly appreciate if you could consider this information and send your nominations till 22 December, 2022.

On behalf of the MJC RUD I thank you for your consideration, and look forward to hearing your nominations.

Yours,



Béla Melegh  
MJC RUD President

Prof. Borut Peterlin  
President of ESHG

15 November, 2022

Dear Mr. President,  
Dear Borut,

I'm contacting you on behalf of the Multidisciplinary Joint Committee of Rare and Undiagnosed Diseases (MJC RUD) of the European Union of Medical Specialties (UEMS). Here I would like to initiate a discussion in two related, but still separate issues.

The first is to initiate a partnership in the MJC RUD system of European assessment in rare and undiagnosed diseases, which is under development, with an ultimate goal of launch a "European Exam in Essentials of Rare and Undiagnosed Diseases". Luckily, I do not have to go too much into the details, since you and the ESHG Exec members are familiar with the UEMS European Exams: we already collaborated in the launch of European Exam in Medical Genetics and Genomics, which was introduced under my presidency of Section of Medical Genetics (SMG) in 2018. In this sequence the ESHG could remain a crucial partner of us. The prestige of the certificate was hugely increased by the fact that the ESHG logo is on the diploma, and the statement "This certificate is recognized as specialist certification in medical genetics by the European Society of Human Genetics" is printed on it (copy of original is attached). Essentially this is the same partnership we propose now for the examination in RUD. To solve the possible worries and concerns, I have to underline here again, that the participation of the ESHG would not include any liability consequence, as it wasn't there in the SMG exam as well.

The core rare diseases exam would be something new. At the moment no such qualification exists in any country, but in some nations authorities do work on it, like Australia, UK, and Lithuania. In our exam a quite prestigious exam committee already supports the goals, like Domenica Taruscio, Wendy van Zelst Stams, Bruce Korf, William Gahl, Gareth Baynam, are prominent names from the long list.

The second issue is also related to the rare disease tasks. At the UEMS Council of 29-30. April of 2022, creation of the European Board of Rare and Undiagnosed Diseases (EBRUD) was approved, which means in UEMS terms that this approval authorized the MJC RUD to proceed with the organization of the EBRUD, according to the RoP of the UEMS. The UEMS European Boards are working groups of the UEMS Bodies but it is of utmost importance and value that recognized academic or scientific societies participate in their work by delegating representatives. Due to the nature of the current and future MJC RUD activity, presence of the ESHG would be pivotal.

On behalf of the MJC RUD I thank you for your consideration, and look forward to hearing your decision.

Yours,



Béla Melegh  
MJC RUD President



EUROPEAN UNION of MEDICAL SPECIALISTS  
Specialist Section of Laboratory Medicine/Medical Biopathology

[www.uems-slm.org](http://www.uems-slm.org)

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18.11.2022

Dear Bela,

The Section of Laboratory Medicine noted that on the agenda of the online Annual Meeting of MJC RUD 15 November, 2022 the topic of

“WG for Laboratory Genetics, ETR for Clinical Laboratory Genetics - proposal for creation this new WG, with the long-term goal of construction of an ETR”

Laboratory Genetics has been part of the ETR for Laboratory Medicine since its inception. The Section of Laboratory Medicine further noted that the Section has not received any information regarding this proposal from MJC RUD despite this long history. The unilateral proposal of the creation of a separate ETR for Clinical Laboratory Genetics goes against this long history of the Section of Laboratory Medicine's ETR.

Laboratory Genetics continues to be heavily grounded in Laboratory Medicine and this was unanimously agreed by our Section delegates as recently as 5th November 2022. Although no concerns have been expressed to us that the existence of the ETRs for Laboratory Medicine and for other specialities could potentially cause confusion in individual countries, (since our last ETR was approved) there has been on our initiative, increased co-operation between the Section of Laboratory Medicine and other Sections concerning areas of mutual interest. I am happy to report that there is a constructive and collaborative working relationship between the Section of Laboratory Medicine and other Sections. I have no doubt that this will continue as our respective ETRs are updated.

The creation of an extra ETR under the MJC RUD does not seem to be necessary. More importantly, this has the potential to cause confusion in individual countries especially concerning the role of medical specialists in Laboratory Genetics.

As stated at the MJC RUD 15 November meeting, we noted that this topic is not urgent and were grateful that you acquiesced to our request to postpone this topic from the agenda with a view to discussing this topic offline. In advance of any such meeting, I would be grateful if you could pass on relevant information to myself and relevant stakeholders copied in so that we can prepare for the meeting in a colligate fashion.

Yours sincerely

Stylianos Chatzipanagiotou  
Section Secretary

Patrick Twomey  
Section President