

## PROGRAM: THURSDAY 28 FEBRUARY

8.00	Registration open	
08.30-10.30	<b>SESSION 1: State of the art lectures</b>	Chair: Ida SCHWARTZ
	Opening address:	Carlo DIONISI-VICI Tara MORRISON
	Metabolism	
8.30 – 8.50	Sulfur Amino Acids	Viktor KOZICH
8.50 – 9.10	Folate and B12	Henk BLOM
	Clinical Presentations	
9.10 – 9.30	CBS deficiency: Overview	Sufin YAP
9.30 – 9.40	CBS deficiency: A parents' perspective	Tara MORRISON
9.40 – 10.00	CblC and MTHFR: Overview	Carlo DIONISI-VICI
10.00 – 10.10	CblC: an adults' perspective	Francesca RESTUCCIA
10.10	<b>COFFEE BREAK</b>	
10.30-11.10	<b>SESSION 2: Novel Developments in Remethylation Defects</b>	Chair: Henk BLOM
10.30 -10.50	A transgenerational MMACHC epimutation illustrates a new mechanism of inborn errors of metabolism named epi-cblC	Jean-Louise GUEANT
10.50 -11.10	Structural basis of severe 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency	Matthias BAUMGARTNER
11.10 – 13.10	<b>SESSION 3: Newborn screening</b>	Chair: Alberto BURLINA
11.10 – 11.40	EHOD Recommendations on newborn screening for homocystinurias versus current practices	Martina HUEMER
11.40 – 12.00	Technical aspects of NBS in Homocystinurias	Giancarlo LA MARCA
12.00 – 12.20	Clinical management of a positive NBS for Homocystinurias	Alberto BURLINA
12.20 – 12.50	The outcomes in classical homocystinuria patients detected by newborn screening	Tawfeg BEN-OMRAN
12.50 – 13.10	Novel Developments in Remethylation Defects and Newborn screening: Q & A	Session speakers
13.10	<b>LUNCH</b>	
14.00 – 15.30	<b>SESSION 4: Interactive Panel</b>	Chair: Kimberly CHAPMAN
14.00 – 14.45	CBS deficiency	Viktor KOZICH Tawfeg BEN-OMRAN Sufin YAP
14.45 – 15.30	Remethylation disorders	Martina HUEMER Carlo DIONISI-VICI Matthias BAUMGARTNER Irina MANOLI
15.30	<b>COFFEE BREAK</b>	
	<b>SESSION 5: New Developments</b>	Chair: Viktor KOZICH
15.50 – 16.10	Microbiome in CBS deficiency	Ida SCHWARTZ
16.10 – 16.30	Arg to Cys mutation in CBS deficiency	Henk BLOM
16.30 – 16.45	Closing comments: Day 1	Henk BLOM Ida SCHWARTZ
17.00 – 18.00	<b>NETWORKING CANAPES &amp; DRINKS</b>	



## PROGRAM: FRIDAY 1 MARCH

8.00	Registration open	
08.30-10.30	<b>SESSION 6: Long term outcomes</b>	Chair: Carlo DIONISI-VICI
8.30 - 9.00	E-HOD Registry results: Remethylation disorders	Martina HUEMER
	E-HOD Registry results: CBS deficiency	Viktor KOZICH
9.00 - 9.20	The Irish Experience	Gregory PASTORES
9.20 - 9.40	Observational study of the natural history of patients with classical homocystinuria on current therapy	Marcia SELLOS-MOURA
9.40 - 11.00	<b>SESSION 7: Recent advances in novel treatments</b>	Chair: TBC
9.40 - 10.00	Enzyme replacement therapy for classical homocystinuria	Tomas MAJTAN
10.00 - 10.20	Impact of EPI743 on visual function in cblC defect	Diego MARTINELLI
10.20 - 10.40	Progress in CblC treatment	Irini MANOLI
10.40 - 10.50	Treatment of cobalamin metabolic disorders with oh-cobalamin through an "in situ" subcutaneous permanent catheter: reaching the goal.	Andrea BORDUGO
10.50 - 11.00	Long term outcomes and novel treatments: Q & A	Session Speakers
11.00	COFFEE BREAK	
11.20 - 13.00	<b>SESSION 8: Living with homocystinuria</b>	Chair: Ida SCHWARTZ
11.20 - 12.00	Dietary practices in pyridoxine non-responsive homocystinuria and the impact of E-HOD guidelines	Marjorie DIXON Alexandra JUNG
12.00 - 12.15	RMD calculator& platform- tools supporting HCU diet calculation and communication between HCP & patients	Iwona BARTLOMIEJCZYK
12.15 - 12.25	Classical Homocystinuria in Qatar: Nutritional advances and challenges	Reem AL-SAAD
12.25 - 12.40	The challenges of parenting a child with phenylketonuria (PKU): Parents' lived experiences	Anja WITTKOWSKI
12.40 - 12.45	Growing up with classical homocystinuria: a child's perspective.	Owen AUSTIN
12.45 - 13.00	Living with homocystinuria: Q & A	Session speakers
13.00	LUNCH	
14.00 - 15.30	<b>SESSION 9: Patient Organisations</b>	Chair: Tara MORRISON
14.00 - 14.20	Global Research Map and Grants Process for HCU	Margie MCGLYNN
14.20 - 14.40	Homocystinuria Patient and Caregiver Survey: Experiences of Diagnosis and Patient Satisfaction	Tara MORRISON
14.40 - 15.00	Cblc onlus, from the beginning until now	Rossella BRINDISI
15.00 - 15.20	cblC Brazil - News and Progress	Luana BRITO
15.20 - 15.30	Closing comments: Day 2	Henk BLOM Ida SCHWARTZ
	Acknowledgements	Carlo DIONISI-VICI Tara MORRISON
15.30	CLOSE	

