5. European Rett Syndrome Congress

02.-04. November 2017 | Berlin

to connect people – to share knowledge

Date	Time		Sessions	Sessions	Workshops
Th, 02.11.	11:00 – 12:30	Opening ceremony	**Welcome** Opening session President of Rett Elternhilfe, Gaby Kessler Congress President, Professor B. Wilken Chairs: F. Hanefeld , G. Kessler Rett Syndrome, the rise and natural history *A. Percy (Birmingham, USA)* Reading from her book ”Brief an meine Schwester“ *L. Malton (Berlin, Germany)*		
	12:30 – 14:00	Break			
	14:00 – 15:30		**Session 2** **MeCP2 function** Chairs: P. Burfeind, P. Huppke 14:00 – 14:30 Role of the MeCP2 protein in activity-dependent neuronal responses *L. Boxer (Boston, USA)* 14:30 – 15:00	**Session 3** **Communication** Chairs: T. Bartolotta, J. Lunding 14:00 – 14:20 Linguistic´s of speech initiation and support *K. Nonn (Munich, Germany)* 14:20 – 14:40 Communication in individuals with Rett Syndrome	**AKUK 1** 14:00 – 15:30 Rett-Basic *M. Westphal (Barnstedt, Germany) and A. von Lukas-Sterner (Rostock, Germany)*

			MECP2 expression and Rett Syndrome-like phenotypes *P. Ross (Glasgow, UK)* 15:00 – 15:30 MeCP2 and regulation of cortical plasticity *S. Shea (Cold Spring Harbor, USA)*	*G. Townend (Maastricht, Netherlands)* 14:40 – 15:00 Communication strategies for non-speaking children, core and fringe Vocabulary *J. Boenisch (Cologne, Germany)* 15:00 – 15:20 Transfer of communication in daily life *J. Wine (Karkur, Israel)* 15:20 – 15:30 general discussion	
	15:30 – 16:00	Break			
	16:00 – 18:00		**<u>The World Café:</u>** <u>Chairs:</u> D. Maack, H.J. Christen *T. Bast (Kork, Germany) and* *B. Wilken (Kassel, Germany):* Epilepsy *L. Malton (Berlin, Germany) and* *H. Leonard (Subiaco, Australia):* Transition *J.M. Ramirez (Seattle, USA) and* *J. Turk (London, United Kingdom):* Breathing disturbance, sleeping problems *G. Townend (Maastrich, Netherlands), H. Wandin (Uppsala, Sweden) and T. Bartolotta (West Long Branch, USA):* International guidelines for communication Concluding remarks by D. Maack and H.J. Christen Plenum 17.45-18:00	**<u>Session 4:</u>** **Free papers: Challenges in genetics research** <u>Chairs:</u> F. Laccone (Vienna, Austria) 16:00 – 16:10 A novel Mecp2 Y120D knockin mouse model manifests typical “Rett-like” behavioral phenotypes but distinct molecular features with respect to the null background *N. Landsberger (Milano, Italy)* 16:10 – 16:20 Primary cilia require a functional MeCP2: can MECP2 patients be affected also by a ciliopathy? *N. Landsberger (Milano, Italy)* 16:20 – 16:30 Genotype-phenotype databases for Rett syndrome and where to find them *F. Ehrhart (Maastricht, Netherlands)* 16:30 – 16:40	**<u>Orthopedic devices</u>** 16:00 – 18:00 *S. Morth (Kassel, Germany) and M. Hoffmann (Kassel, Germany)* **<u>AKUK 2</u>** 16:00 – 18:00 Talker structures / UK ideas and implementation in practice *C. Hirsch (Amelinghausen, Germany) and* *M. Rosenits (Eberbach, Germany)*

				Reduced responsiveness to external stimuli affects developing Mecp2 null neurons and may represent a target for future therapeutic approaches *L. Scaramuzzaa (Milano, Italy)* 16:40 – 16:50 Mechanisms of transcriptional regulation by MeCP2 *J. Cholewa-Waclaw (Edinburgh, United Kingdom)* 16:50 – 17:00 The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome *S. Vidal (Barcelona, Spain)* 17:00 – 17:10 Radically shortened MeCP2 reverses Rett syndrome-like neurological defects *J. Selfridge (Edinburgh, United Kingdom)* 17:10 – 17:20 Clinical and genetic investigation of a Danish cohort of 35 patients with Rett-like phenotypes *B. Schönewolf-Greulich (Glostrup, Denmark)* 17:20 – 17:30 general discussion	
	18:00 – 21:00		Get Together		Andel`s Hotel
Fr, 03.11.	9:00 – 11:00		**<u>Session 5</u>** **Basic research** <u>Chairs:</u> P. Huppke, P. Burfeind 09:00 – 09:30 Characterization of autonomic dysfunction in Rett Syndrome and other MeCP2 disorders *J. Neul (Nashville, USA)*	**<u>Session 6</u>** **Special issues in Rett Syndrome** <u>Chairs:</u> B. Wilken, J.-M. Ramirez 09:00 – 09:30 Sleep disturbances in Rett Syndrome *J. Turk (London, United Kingdom)*	**<u>Communication</u>** 9:00 – 11:00 Developing ways of using AAC – illustrated by the example of a girl with Rett Syndrome

			09:30 – 09:50 Role of MeCP2 in regulating chromatin organization/remodeling *C. Cardoso (Darmstadt, Germany)* 09:50 – 10:10 Genomics of Rett Syndrome *M. Esteller (Barcelona, Spain)* 10:10 – 10:30 Physiological and pathophysiological role of MeCP2 expression on central synapse function and synapse formation *C. Rosenmund (Berlin, Germany)* 10:30 – 10:50 MeCP2 deficiency, microtubule dynamics and vesicular transport *T. Bienvenu (Paris, France)* 10:50 – 11:00 general discussion	09:30 – 10:00 Breathing abnormalities and regulation in Rett *J.M. Ramirez (Seattle, USA)* 10:00 – 10:30 Vibration training physiotherapy (Galileo) in Rett Syndrome *E. Schönau (Köln, Germany)* 10:30 – 11:00 Bone health in Rett Syndrome *E. Schönau (Cologne, Germany)*	*C. Dieckmann (Fuerth, Germany)*
	11:00 – 11:30	Break			
	11:30 – 12:30		**<u>Session 7</u>** **Transition** <u>Chairs:</u> A. Ferbert , W. Asthalter 11:30 – 12:00 Transition in Rett Syndrome *H. Leonard (Subiaco, Australia)* 12:00 – 12:30 Aging in Rett Syndrome *M. Freilinger (Vienna, Austria) and*	**<u>Session 8</u>** **Free papers: free communications** <u>Chairs:</u> M. Müller 11:30 – 11:40 Abnormal foot position and standing and walking ability in Rett syndrome: an exploratory study *H. Borst (Maastricht, Netherlands)* 11:40 – 11:50	

			E. Smeets (Maastricht, Netherlands)	Eye Tracking devices and cognitive capacity in Rett Syndrome: a research study *G. Martino (Messina, Italy)* 11:50 – 12:00 Eye-gaze technology as a communication device: habilitation process with three subjects with Rett Syndrome *L. Lintula (Tampere, Finland)* 12:00 – 12:10 The Preliminary Study of AAC Service Delivery Model for Children with Rett syndrome in China *T.W. Wang (Taipei, Taiwan)* 12:10 – 12:30 general discussion	
	12:30 – 14:00	Lunch (12:30 – 13:30)	**<u>Country Updates – Room "Opal"</u>** 12:30 – 14:00 **<u>Poster walk 1:</u>** 13:00 – 13:30 <u>Chair:</u> S. Schulze Poster 1-6		
	14:00 – 15:30		**<u>Session 9</u>** **Communication** <u>Chairs:</u> C. Petzold, G. Townend 14:00 – 14:20 Emergent Literacy for Girls with Rett Syndrome *S. Sachse (Cologne, Germany)* 14:20 – 14:35 Combining AAC and Partner Coaching *T. Bartolotta (West Long Branch, USA)* 14:35 – 14:50	**<u>Session 10</u>** **Free papers** <u>Chairs:</u> S. Hülsmann, W. Asthalter 14:00 – 14:10 Role of inhibitory neurons for the breathing pathology in mouse models of Rett Syndrome *S. Hülsmann (Göttingen, Germany)* 14:10 – 14:20 Cellular redox imbalance and oxidative stress in Rett Syndrome: Deciphering their roles as potential key players in disease progression	

			Visual attention when aided language modeling is used in interaction *H. Wandin (Uppsala, Sweden)* 14:50 – 15:05 Realtime video registration of communication through eye gaze devices in girls with Rett Syndrome *G. Scholte (Utrecht, Netherlands)* 15:05 – 15:15 general discussion	*M. Müller (Göttingen, Germany)* 14:20 – 14:30 The 5-ht5b receptor interacts with other receptor subtypes - implications for signaling defects in Rett Syndrome *M. Niebert (Göttingen, Germany)* 14:30 – 14:40 Epilepsy in Rett syndrome from a lifetime perspective *M.W.Henriksen (Drammen, Norway)* 14:40 – 14:50 Non epileptic paroxysmal events in Rett Syndrome *A. Vignoli et al. (Milano, Italy)* 14:50 – 15:00 Management of Emotional, Behavioural and Autonomic Dysregulation (EBAD) in patients with Rett Syndrome *P. Santosh (London, United Kingdom)* 15:00 – 15:10 Aberrant glutamate handling and signaling in Rett mouse hippocampus *S. Mironov (Göttingen, Germany) and* *S. Balakrishnan (Göttingen, Germany)* 15:10 – 15:20 Rett syndrome spectrum-like phenotype in siblings with ALG1-CDG *A.T. Midro et al. (Białystok, Poland)* 15:20 – 15:30 general discussion	
	15:30 – 16:00	Break			
	16:00 – 17:30		**<u>Session 11</u>** **CDKL5**	**<u>Session 12</u>** **Orthopedic and related Problems**	**<u>Communication</u>** 16:00 – 17:30

			Chairs: A. Panzer, C. Fuchs 16:00 – 16:20 The molecular mechanisms of CDKL5 *T. Pizzorusso (Florence, Italy)* 16:20 – 16:40 Experimental therapeutic approach for CDKL5 *C. Fuchs (Bologna, Italy)* 16:40 – 17:00 Phenotypes and the therapeutic challenges of CDKL5 *S. Philip (Birmingham, United Kingdom)* 17:00 – 17:20 Combination of intensive physiotherapy in treatment of children with CDKL5. The Footsteps Center *A. Mason (Wallingford, United Kingdom)* 17:20 – 17:30 general discussion	Chairs: A.-K. Hell, J. Downs 16:00 – 16:30 Experience with Transition, a single center experience *A. Ferbert (Kassel, Germany)* 16:30 – 17:00 Motor development *J. Downs (Subiaco, Australia)* 17:00 – 17:30 Scoliosis in Rett Syndrome *A.-K. Hell (Göttingen, Germany)*	"Rett-CHAT" - Communication - Hope - Again - Technology *R. Malzer (Pichl, Austria)*
	17:30 – 17:45	Break			
	17:45 – 18:45		**Session 13** **Round table discussion: Direction of future research projects** Moderation: P. Huppke Participants: M. Freilinger (Vienna, Austria) G. Townend (Maastricht, Netherlands) J. Neul (Nashville, USA) W. Kaufmann (Boston, USA) P. Burfeind (Göttingen, Germany) L. Boxer (Boston, USA)		Shuttle Kosmos: 19h20 / 19h30

	19:30		Social evening		
Sa, 04.11.	08:30 – 09:30		**Poster walk 2:** 08:30 – 09:20 Chair: A. Panzer Poster 7-16		
	09:30 – 11:00		**Session 14** **Epilepsy** Chairs: T. Bast, H.J. Christen 09:30 – 10:00 Clinical manifestation and prognosis *T. Bast (Kork, Germany)* 10:00 – 10:30 Antiepileptic therapy *A. Vignoli et al.(Milano, Italy)* 10:30 – 11:00 Differential diagnosis of epileptic seizures *T. Dietel (Kork, Germany)*	**Session 15** **Free Papers: Special issues in Rett Syndrome** Chairs: M. Freilinger, E. Smeets 09:30 – 09:40 Growth of Dutch females with Rett Syndrome: Comparison with Healthy Dutch Females and American Females with Rett Syndrome *E. Smeets (Maastricht, Netherlands)* 09:40 – 09:50 Long distance home-based, physical therapy, consultation for individuals with Rett Syndrome – a pilot study *M. Lotan (Ariel, Israel)* 09:50 – 10:00 Findings from a national evaluation of 145 Italian Rett syndrome cohort, over the years 2013-2017 *M. Lotan (Ariel, Israel)* 10:00 – 10:10 Enablers and barriers to 'up-time' activities in girls and women with Rett syndrome - Perspectives from parents and professionals *M. Stahlhut (Copenhagen, Denmark)* 10:10 – 10:20 Physical activity patterns across ambulation levels in girls and women with Rett syndrome *M. Stahlhut (Copenhagen, Denmark)*	**Transition** 9:30 – 11:00 A report from the German Rett Syndrome working group on the successful outcome of their 12 year study on ageing and transition to adulthood The number of participants is restricted to 50 (registration on-site, no early sign-up necessary). Language: English / German W. Asthalter (Koerle, Germany)

				10:20 – 10:30 Establishing a University-Based Center for Research and Support for Individuals with Rett Syndrome and their Families *P.A. Remshifski (West Long Beach, USA)* 10:30 – 10:40 Important symptoms of Rett Syndrome and their impact: the perspective of parents and healthcare providers *K. Williams (London, United Kingdom)* 10:40 – 10:50 Aging in Rett syndrome: risk of decline in gross motor skills *A.-M. Bisgaard (Copenhagen, Denmark)* 10:50 – 11:00 You never walk alone - Gait Training for Children and Teenagers with Rett Syndrome *A. Jagusch-Espei (Münster, Germany)*	
	11:00 – 11:30	Break			
	11:30 – 13:30		**<u>Session 16</u>** **New treatment options** <u>Chairs:</u> W. Kaufmann, F. Laccone 11:30 – 12:00 Treatment options in Rett Syndrome, opportunities and false hopes *W. Kaufmann (Boston, USA)* 12.00 – 12:20 Update protein replacement therapy *F. Laccone (Vienna, Austria)* 12:20 – 12:40 Update MeCP2 reactivation		

			J. Gribnau (Leiden, Netherlands) 12:40 – 13:00 Update Gene therapy in Rett Syndrome *K. Gadalla (Glasgow, UK)* 13:00 – 13:20 Genome editing using CRISPR-Cas9 - Applications in Rett Syndrome and neurodevelopmental disorders *A. Banerjee (Zürich, Switzerland)* 13:20 – 13:30 general discussion		
	14:00		**Farewell and concluding remarks** B. Wilken, G. Kessler End of congress		

Poster - walk 1: Fri. Nov. 3rd –13:00 -13:30

1. L. Hinz (Amsterdam, Netherlands): *Generation of isogenic controls for in vitro disease modelling of X-chromosome-linked disorders*

2. N.B. Sangani (Maastricht, Netherlands): *Protein interaction analysis of HDAC6, a target of MeCP2 with therapeutic interest for Rett syndrome*

3. S. Bettini (Bologna, Italy): *Characterization of CDKL5 disorder-like phenotypes in heterozygous Cdkl5 KO female mice*

4. G. Medici (Bologna, Italy): *SMAD3, a novel CDKL5 substrate, plays a role in the defective survival and maturation of Cdkl5-null neurons*

5. D. Peroni (Busto Arsizio, Italy): *Therapeutic relevance of pregnenolone-based approaches for disorders linked to CDKL5-deficiency*

6. M. Tamarin (Busto Arsizio, Italy): *CDKL5 associated AMPA-receptor defects: a novel target for drug-based therapies?*

Poster - walk 2: Sa. Nov. 4th – 8:30 – 9:30

7. O.H. Skjeldal (Gothenburg/Drammen, Sweden/Norway): *Classical Rett syndrome in a patient with a de novo mutation in SCN1a*

8. G. Prato et al. (Genoa, Italy): *Cannabinoids experience in three patients with CDKL5 and MECP2 mutations*

9. C. Miranda-Lourenço et al. (Lisbon, Portugal): *Adenosinergic system as a new central player in Rett Syndrome*

10. J.L. Larsen (Copenhagen, Denmark): *What do parents to girls and women with Rett syndrome worry about?*

11. D. Schwab (Grenoble, France): *GazePlay: Creation of a community to help the development of a Free and Open-source plateform to make eye-tracker Video Games accessible to everyone.*

12. J. Mathieu (Munich, Germany): *Approaches to emergent literacy support for girls with Rett-Syndrome in speech language therapy – a proposal*

13. S.M. Graham et al. (Morristown, USA): *Sarizotan Treatment of Severe Apnoea in Rett Syndrome (STARS) - from Concept to Clinical Trial: Design of a Global, Double-Blind, Placebo-Controlled Study*

14. C. Kronborg (Copenhagen, Denmark): *Do Danish girls/women with Rett syndrome meet the international scoliosis guidelines?*

15. C. Kronborg (Copenhagen, Denmark): *StepWatchTM Activity Monitor versus Yamax Digi-Walker® in Rett syndrome – a pilot study*

16. F. Camia (Genova, Italy): *AIRett Clinical Passport: a safe solution for medical management*

17. J. Guy (Edinburgh, UK): *Rett Mutations outside two key functional domains of MeCP2 affect protein abundance and corepressor recruitment*

18. S. Russo (Milano, Italy): *Generation of Rett patient-specific iPSCs (reprogrammed from blood) for biomarker discovery and drug screening*