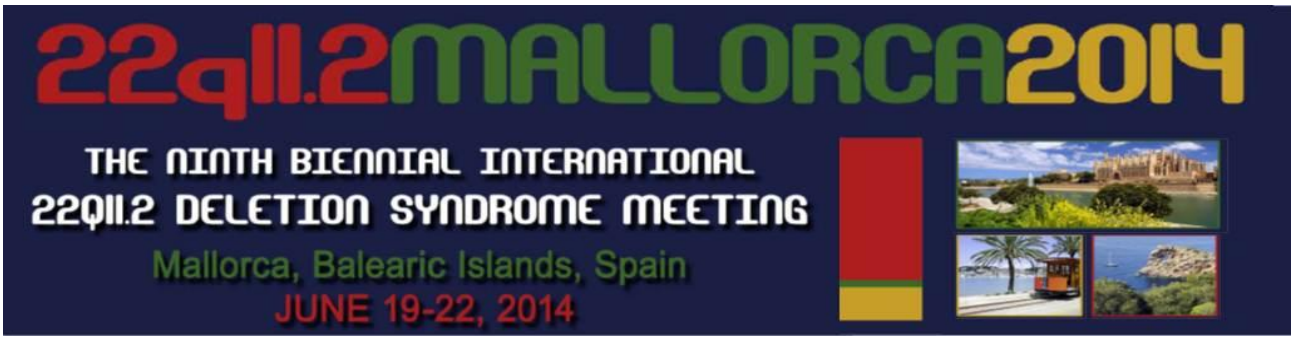

PROFESSIONAL PROGRAM

THURSDAY JUNE, 19TH (7:30-19:30h)

7:30	**Conference registration begins**
8:30	**Welcome and conference opening**
9:00-11:00	**BIOLOGY OF THE SYNDROME: Model organisms**
9:00-9:45	Invited presentation : **Francesco Papaleo**- Studying cognitive development and schizophrenia-like phenotypes in genetically modified mice relevant to 22q11DS
9:45-10:10	**Peter Scambler-**Investigation of the CXCL12-CXCR4 pathway in cardiovascular morphogenesis and its possible role downstream of TBX1
10:10-10:35	**Silvia Racedo-**Reduction of β-catenin gene dosage in the secondary heart field mesoderm rescues TBX1 mutant cardiac phenotype
10:35-11:00	**Anthony Lamantia-**Why are feeding and swallowing disrupted in 22q11 Deletion Syndrome?
11:00-11:30	**Coffee break**
11:30-13:30	**BIOLOGY OF THE SYNDROME: Patients**
11:30-12:00	Invited presentation : **Title to be determined**
12:00-12:15	**Beverly Emmanuel-**A duplication of *SLC2A3* modifies the cardiovascular phenotype in 22q11DS
12:15-12:25	**Loydie Jerome-Majewska-**Hemizygous Mutations in SNAP29 and SCARF2 Contribute to Atypical Findings in Patients with 22q11.2DS
12:25-12:35	**Mathew Hestand-**A catalog of hemizygous variation in 127 22q11 deletion patients
12:35-12:45	**Flora Tassone-**Molecular underpinning of the 22q11 deletion syndrome: Genotype/Phenotype correlations
12:45-12:55	**Damian Heine-**Characterization of a family with two siblings carrying de novo paternally derived deletion syndromes (22q11.2 and 17q21.31).
12:55-13:05	**Alexander Urban-**Genomic characterization and copy number analysis of human induced pluripotent stem cells (iPSCs) from 22q11.2 deletion syndrome patients.
13:05-13:15	**Weng Zhang-**Identification of causal genes of tetralogy of Fallot in 22q11DS using sequencing-based logistic regression with network regularization
13:15-13:25	**Aaron Golden-**The International Brain and Behavior Consortium Genomics Gateway
13:30-15:00	**Lunch and POSTER SESSION I**

	MEDICAL ASPECTS AND HEALTHCARE
	Genotype-Phenotype
15:00-15:45	**Invited presentation: Luis Perez-Jurado:** Comparison Williams and 22q11.2 Syndromes
15:45-16:00	**Donna McDonald-McGinn-**The perplexing prevalence of familial nested 22q11.2 deletions
16:00-16:15	**Maria Digilio-**Clinical characteristics of patients with deletion 22Q11.2 and atypical size
16:15-16:30	**Gabriela Repetto-**22Q11 deletion size in Chilean patients and association with clinical features
16:30-17:00	**Coffee break**
17:00-18:30	**MEDICAL ASPECTS AND HEALTHCARE**
	Screening and diagnosis
17:00-17:30	**Invited Presentation: Title to be determined**
17:15-17:30	**Jenny Lingman Framme-**Retrospective analysis of TREC based newborn screening results and clinical phenotypes in infants with the 22q11 deletion syndrome.
17:30-17:45	**Gozde Akzumus-**Congenital diaphragmatic hernia as an important clue to the diagnosis of 22q11.2 Deletion Syndrome
17:45-18:30	**Invited presentation-Eduard Gratacos:** In utero treatment of DiGeorge related malformations
18:30-19:30	**Wine aperitif and POSTER SESSION II**
21:00	**Gala Dinner**

FRIDAY JUNE, 20TH (8-19h)

8:00-10:10	**MEDICAL ASPECTS AND HEALTHCARE**
	Heart
9:00-9:10	**Derk Verschure-**Cardiac sympathetic activity in 22q11 deletion syndrome: outline of proof of concept study
	Hearing
9:10-9:20	**Greet Hens-**High prevalence of sensorineural hearing loss in 22q11.2 Deletion Syndrome
	Gastrointestinal
9:20-9:30	**Kesav Anupindi-**Assesment of upper gastrointestinal exams in children with 22q11.2 Deletion Syndrome
	Overgrowth-Duplications
9:30-9:40	**Elaine Zackai**-Overgrowth as a presenting symptom of the 22q11.2 Duplication Syndrome – A novel association
	Sleep
9:40-9:50	**Pamela Mudd**-22q11.2 Deletion Syndrome and obstructive sleep apnea
	Velopharyngeal dysfunction
9:50-10:00	**Kaitlyn Paine**-Practice patterns for management of velopharyngeal dysfunction in patients with 22q11.2 Deletion Syndrome
	Scoliosis
10:00-10:10	**Dino Colo-**Scoliosis in the 22q11.2-deletion syndrome
10:10-10:40	**Coffee break**

10:40-12:20	**MEDICAL ASPECTS AND HEALTHCARE** **Adults**
10:40-11:05	**Ann Swillen-**Presenting symptoms in adults with the 22q11 Deletion Syndrome
11:05-11:15	**Erik Boot-**Movement and gait abnormalities in adults with 22q11.2 Deletion Syndrome
11:15-11:25	**Christina Persson-**Speech in adults with 22q11.2 Deletion Syndrome: A pilot study
11:25-11:35	**Nicole Spruijt-**Self-reported speech problems in adolescents and young adults with 22q11.2 Deletion Syndrome: A retrospective study
11:35-11:45	**Marta Unolt-**Clinical follow-up of adolescent/adult patients with 22q11.2 Deletion Syndrome: Preliminary data
	Brain
11:45-12:10	**Anne Bassett-**22q11.2 deletion syndrome, intrauterine growth and neurodevelopmental outcome
12:10-12:20	**Paula Goldenberg-**Higher frequency of polymicrogyria in a cardiovascular cohort with 22q11.2 deletion syndrome
12:20-13:00	**EDUCATION**
12:20-12:30	**Susan Busch-**Lessons Learned
12:30-12:40	**Alan Fung-**Development of a support group for adults with 22q11.2 deletion syndrome
12:40-12:50	**Stephen Hooper-**Social Cognitive Training (SCT) for Adolescents with 22q11: Preliminary Findings
12:50-13:00	**Linda Campbell-**A cultural perspective of diagnosis: A study of parents' experiences of their child's diagnosis of 22q11DS
13:00-14:30	**Lunch and POSTER SESSION III**
14:30-18:30	**BRAIN AND BEHAVIOUR**
14:30-14:55	**Jacob Vortsman-**Is early cognitive decline associated with the risk of psychotic illness in patients with the 22q11.2 deletion syndrome? A study by the International 22q11.2 Brain Behavior Syndrome Consortium.
14:55-15:05	**Raquel Gur-**Subthreshold psychotic symptoms in 22q11.2 Deletion Syndrome
15:05-15:15	**Maria Niarchou-**Comparing autism spectrum disorders, psychotic experiences and cognition in children with 22q11.2 Deletion Syndrome with children with 16p11.2 deletions and duplications
15:15-15:25	**Angers Vangkilde-**A neuropsychiatric profile of 279 Danes with 22q11 deletion syndrome - A unique study based on the Danish National Health Registers.
15:25-15:35	**Vandana Shashi-**Deviant Longitudinal Trajectory of Neurodevelopment in Children and Adolescents with 22q11DS
15:35-15:45	**Stephan Eliez**- Atypical developmental trajectories of cognitive functioning and social skills in 22q11.2 deletion syndrome: clinical and cerebral correlates
15:45-15:55	**James Yi-**Association of psychiatric burden and cognitive domains to functional outcome in 22q11.2 Deletion Syndrome
15:55-16:05	**Samuel Chawner-**Longitudinal study indicates 22q11.2 Deletion Syndrome is associated with developmental deficits across multiple neurocongnitive domains in early adolescence
16:05-16:30	**Coffee break**
16:30-16:40	**Maude Schneider-** Does impaired multitasking underlie the development of negative symptoms in adolescents with 22q11.2 deletion syndrome? A study based on a naturalistic experimental paradigm

16:40-16:50 **Therese Von Amelsvoort-**Plasma amino acids and cognitive function in 22q11 Deletion Syndrome

16:50-17:00 **Ruben Gur-**Structural and functional brain abnormalities in 22q11.2 Deletion Syndrome

17:00-17:10 **Carrie Bearden**-Structural and functional neural dysconnectivity as a predictor of psychotic symptom expression in 22q11.2 Deletion Syndrome

17:10-17:20 **Wendy Kates**-White matter microstructural abnormalities of the cingulum bundle in youth with 22q11.2 Deletion Syndrome: Associations with medication and prodromal symptoms of psychosis

17:20-17:30 **Elisa Scariati**-Organization and structure of the functional brain network in 22q11 Deletion Syndrome

17:30-17:40 **Sasja Duijff**-Explaining the variable penetrance of CNVs: educational background of parents modulate the impact of the 22q11.2 deletion on cognitive functioning

17:40-17:50 **Nancy Butcher**-Antipsychotic treatment with clozapine in adults with 22q11.2 deletion syndrome: Towards a model of personalized care

17:50-18:00 **Abbie Popa-**Timecourse of response to threat stimuli in children with 22q11.2 Deletion Syndrome informs understanding of anxiety

18:00-18:10 **Brenda Finucane**-Developmental Brain Dysfunction: Implications for the Study of Phenotypic Heterogeneity in 22q11.2 Deletions and Duplications

18:10-18:20 **Tonia Rihs-**Overview of high-density EEG with the Geneva 22q11.2 deletion cohort

18:20-18:30 **Eva Chow-**Establishing consensus psychosis group status for the International Consortium on Brain and Behavior in 22q11.2 Deletion Syndrome

18:30 Closure

POSTERS

1 **Silvia Racedo**-Mouse and Human CRKL is dosage sensitive for heart morphogenesis

2 **Paola Ariganello**-Clinical phenotype and regulatory T-cells in 22q11.2 Deletion Syndrome

3 **Afnan Salaka**-Investigating the two-hit hypothesis in the 22q11 Deletion Syndrome

4 **Maria Carmela Padula-**Reduced structural connectivity in the default mode network in 22q11.2 Deletion Syndrome

5 **Miralena Tomescu-**Deviant dynamics of EEG resting state pattern in 22q11.2 deletion syndrome adolescents: a vulnerability marker of schizophrenia?

6 **Juan Bregante**-20 years with 22q11DS patients in Mallorca: the experience of the Cleft Unit

7 **Catalina Cancrini-**Clinical features of a large cohort of 22q.11 del 22 syndrome: an Italian multicenter study

8 **Brian Forbes**-Ocular Findings associated with Chromosome 22q11.2 Duplication

9 **Damia Heine Suñer-**Ten years of prenatal screening for 22q11.2 deletions and duplications

10 **Yulia Kozlova**-22q11.2DS Phenocopies Show Etiological Heterogeneity

11 **Aebele Mink van der Molen**-Platybasia in 22q11.2 Deletion Syndrome is not correlated with the speech resonance

12 **Fernando Santos Simarro-**Co-occurrence of 22q11 and 17q12 mosaic microdeletions in a single patient

13 **Elfi Vergaelen**-A case report on a 3 generation family with paternal transmission of the 22q11.2 Deletion Syndrome: Intrafamilial phenotypic variability
14 **Jaume Enjuanes-Llovet**-Chromosome 22q11.2 deletion syndrome: epidemiologic and phenotypic review on 50 Spanish patients
15 **Rosella Capollino**-Congenital heart defects in the 22q11.2 distal deletion: Noncompaction of the left ventricle with aortic valve anomalies
16 **Elisa Chioma**-Neuropsychological manifestations in patients with 22q11.2 Deletion Syndrome
17 **Joseph F Cubells-**Examining the Overlap of Autism Spectrum Disorder and 22q11 Deletion Syndrome Using Standardized Clinical Assessments
18 **Anna Mª Cueto Gonzalez**-Atypical 22q11.2 deletion

19 **Diego Desotto-**22q11 Deletion Syndrome : review of our experience in a tertiary hospital.
20 **Chiara Squarcione**-Psychopathology in a Sample of 22q11 Deletion Syndrome Italian Adult Patients: a Preliminary Descriptive Study

21 **Luis Fernandez**-22q11.2 deletion syndrome in a clinical setting: a retrospective study
22 **Derya Ercal**-A case with 22q11 deletion associated with hypocalcemic convulsion
23 **María Reyes Jiménez León**-Secondary immunologic consequences in chromosome 22q11.2 Deletion Syndrome in a cohort of 14 patients
24 **Pamela Puliafito**-Immunological alteration in a large cohort of 22q11.2 Deletion Syndrome patients
25 **Elise M Sannar**-Psychopharmacologic Treatment of Anxiety in 22q11.2 Deletion Syndrome: A Case Series
26 **Ellen Van Den Heuvel-**Cross syndrome comparison of referential communication skills: characteristics of children with 22q11.2 Deletion Syndrome

27 **Teresa Vendrell Bayona**-22q11.2 deletion syndrome - haematological abnormalities

28 **Donna Cutler-Landsman**-Bridging the Gap Between Medical Professionals and the Schools