

**XVI Congresso Scientifico Annuale Internazionale
della Velo-Cardio-Facial Syndrome Educational Foundation, Inc.
in collaborazione con Associazione Italiana Delezione Cromosoma 22 Onlus e il
Network Europeo VCFSEF**

3-5 luglio 2009, Roma, Italia

PROGRAMMA

Venerdi mattina, 3 luglio 2009

Iscrizioni: apertura dalle 7:30

9:00 – 9:20: **Cerimonia d'apertura e introduzione**

Karen J. Golding-Kushner (USA), Executive Director, VCFSEF

Giulietta Cafiero (Italia), Amyel Loy (Italia), Bruno Marino (Italia), Aidel22

9:20 – 9:30: **Rassegna storica della SVCF e della VCFS Educational Foundation;**

Robert J. Shprintzen (USA)

9:30 – 10:30: **Moderatore: M. Cristina DiGilio (Italia)**

Aspetti clinici nella Sindrome VCF: Un confronto internazionale

9:30 – 9:40: Manifestazioni cliniche in un gruppo di 48 pazienti con sindrome della delezione 22q11; *Angelo*

Giovanni Delmonaco, Cristina Molinatto, Elisa Biamino, Elga Fabia Belligni, Serena

Forzano, Elena Banaudi, Orsetta Zuffardi, Margherita Silengo, Giovanni Battista Ferrero, Italia

9:40 – 9:50: Aspetti clinici in un gruppo di pazienti con delezione 2q11: uno studio italiano multicentrico;

C. Cancrini, P. Puliafito, E.M. Ruga, S. Martino, C. Putti, A. Soresina, M.C. Di Gilio, R.

Consolini, R. Rondelli, F. Cardinali, G. Corsello, A. Aiuti, C. Pignata, C. Azzari, A. Plebani, P.

Rossi, Italia

9:50 – 10:00: Dati clinici e biologici in un gruppo di 75 pazienti con SVCF; *Pierre Sarda, Francia*

10:00–10:10: Sindrome velo cardio- facciale in Cile; *Gabriela Repetto, Maria Luisa Guzman, Alonso Puga, Juan*

Francisco Calderòn, Carmen Paz Astete, Mariana Aracena, Marta Arriaza, Teresa Aravena,

Patricia Sanz, Cile

10:10 – 10:20: Il trattamento della Sindrome VCF in Giappone; *Toko Hayakawa, Giappone*

10:20 – 10:35: Domande e Discussione

10:35 – 10:50: Pausa Caffè

Moderatore : Pierre Sarda (Francia)

10:50 – 11:05: Cardiopatie: diagnosi e trattamento; *Bruno Marino, A. Carotti, G. Michielon, G. Piacentini, M.*

Cristina Digilio, S. Picardo, R. Di Donato, Bruno Dallapiccola, Italia

11:05 – 11:20: Rilevanza della risposta immune-umorale nella SVCF; *P. Ariganello, S. Di Cesare, P. Puliafito,*

M.L. Romiti, F. Casciano, A. Finocchi, P. Rossi, C. Cancrini, Italia

11:20 – 11:35: Aspetti clinici ed immunologici nella SVCF: studio del follow-up nel primo anno di vita;
M.Capelli, C. Radice, F. Bedetti, A.G. Cimatti, L. Conti, A. Minia, G. Ricci, Guido Cocchi, Italia

11:35 – 11:50: Programmazione delle cure: cosa fare e quando *Anne Marie Higgins, USA*

11:50 –12:00: Domande e Discussione

12:00 – 13:05: **Pranzo con gli esperti**

Aspetti psicologici e psichiatrici nella SVCF
Aspetti legati al linguaggio nella prima infanzia
Aspetti educativi

13:05 – 13:20: Funzione del gene Tbx1: un aggiornamento; *Antonio Baldini, M.D., Italia*

13:20 – 13:35: Modificatori genetici nei pazienti con SVCF e con delezione da 3Mb; *Bernice Morrow; USA*

13:35 – 13:50: Individuazione della SVCF per mezzo di analisi MLPA utilizzando gocce di sangue di neonato essiccate su carta assorbente *K.M. Sørensen, Peter Agergaard, P.N. Andersen, L.A. Larsen, J.P. Schouten, M. Christiansen, Danimarca*

13:50 – 14:05: Delezione 22q11, iperprolinemia, ritardo mentale e psicosi nella SVCF . *Audrey Guilmatre, Solenn Le Gallic, Thierry Frébourg, Dominique Champion, Francia.*

14:05 – 14:15: Domande e discussione

14:15 – 14:30: Pausa Caffé

Moderatore: Dianne Altuna

14:30 – 14:45: Un approccio cognitivo alla terapia logopedica; *Marta Paloscia, Italia*

14:45 – 14:55: Suono, Forma e Colore (SSC) , un metodo di trattamento per i disturbi di linguaggio: un'applicazione clinica dei processi fonologici ; *Ariela Nachmany, Israele.*

14:55 – 15:10: Tecniche logopediche per la SVCF ; *Cheryl L. Lozon, USA*

15:10 – 15:25: Modello di linguaggio olistico per la logopedia nella SVCF ; *Antonio Ysunza, M. D., Sc. D., Ma. Carmen Pamplona, M.A., Messico*

15:25 – 15:55: **Un confronto: Risultati ottenuti nella terapia del linguaggio per la SVCF** *Karen Golding-Kushner, Ph.D. (USA), Eileen Marrinan, M.S., M.P.H. (USA), Cheryl Lozon, (USA), Ariela Nachmany (Israele), Antonio Ysunza (Mexico), Marta Paloscia (Italia)*

15:55 – 16:05: Risultati ottenuti in seguito a palatoplastica o flap faringeo in pazienti con SVCF; *Edward W.Swanson, B.A.S., Stephen R. Sullivan, M.D., Emily B. Ridgway, M.D., Eileen M. Marrinan M.S., M.P.H., John B. Mulliken, MD, USA*

16:05 – 16:15: Pianificazione chirurgica per la ricostruzione della funzione velo-faringea nella SVCF ; *Antonio Ysunza, M. D., Sc. D. Ma. Carmen Pamplona, M.A., Adriana Hernandez, M.D. Fernando Molina, M.D., Messico*

16:15 – 16:30: SVCF: rassegna di 75 casi operati di faringoplastica ; *M. Accordi, S. Agnelli, Italia*

16:30 – 16:50: Il trattamento chirurgico della voce ipernasale è diverso per la SVCF? *Sherard A. Tatum, M.D., USA*

16:50 – 17:00: Domande e discussione

17:00: Assemblea annuale della Fondazione VCFS EF

Sabato mattina, 4 Luglio 2009

9:00 – 16:00: Sessione Poster:

Studio longitudinale sull'evoluzione della percezione del tempo in persone con SVCF ; *Diego de Leon Rodriguez, Svizzera*

Ipotesi di programma di recupero socio-emotivo per individui con sindrome da delezione 22q11.2: uno studio pilota; *M Chabloz, B Glaser, A Lothe, C Pasca, S Eliez, Svizzera*

Lo spettro fenotipico di pazienti di Singapore con SVCF; *Teng Sun Shin, Angeline Lai, Tan Ee Shien, Singapore*

Sistema di sostegno per i pazienti con VCFS in Giappone; *Tomoko Tominaga (Japan)*

Presentazioni

Moderatore: Doron Gothelf (Israele)

9:00 – 9:15: Riconoscimento di forme locali: difficoltà visive nella SVCF ; *Anne Giersch, Bronwyn Glaser, Catherine Pasca, Mélanie Chabloz, Martin Debbané, Stephan Eliez, Svizzera*

9:15 – 9:30: Variazione del percorso dello sguardo in individui con sindrome da delezione 22q11.2 durante il Benton Visual Retention Test; *Marie-Christine Ottet, Bronwyn Glaser, Manolo Fuentes, Catherine Pasca, Mélanie Chabloz, Martin Debbané, Stephan Eliez, Svizzera*

9:30 – 9:50: Una finestra sull'anima: attenzione agli occhi nella SVCF; *Bronwyn Glaser, M. Peelen, Martin Debbané, M. Chabloz, C. Pasca, Stephan Eliez, Svizzera*

9:50 – 10:05: Abilità socio-cognitive e percorso dello sguardo in bambini e giovani con SVCF; *L.E.Campbell, K. McCabe, L. Cruickshank, K. Leadbeater, U. Schall, F. Karayanidis, C.Loughland, Australia*

10:05 – 10:20: Domande e discussione

10:20 – 10:40: Pausa Caffè

Moderatore: Bronwyn Glaser (Svizzera)

10:40 – 10:55: Elaborazione delle emozioni in adolescenti con SVCF: uno studio di risonanza magnetica funzionale; *Ioana Coman, Ph.D., Wendy Kates, Ph.D., Adam Stern, USA*

10:55 – 11:15: Uno studio longitudinale della morfologia e dello spessore corticali nei bambini con SVCF; *Wendy Kates, Ph.D., USA*

11:15 – 11:35: Sviluppo del cervello nella SVCF: uno studio longitudinale; *Marie Schaer, Marie-Christine Ottet, Bronwyn Glaser, Martin Debbané, Stephan Eliez, Svizzera*

11:35 – 11:50: Correlazioni neuroanatomiche delle funzioni del linguaggio e cognitiva nella SVCF; *Eileen Marrinan, M.S., M.P.H., Karen Stroszka, M.S., Robert J. Shprintzen, Ph.D., Kevin Antshel, Ph.D., Wendy Kates, Ph.D., USA*

11:50 – 12:00: Discussione e domande

12:00 – 13:10 **Pranzo con gli esperti:**
Genetica e Genomica
Aspetti del linguaggio in età scolare
Trattamento medico
Alimentazione e crescita

Sabato pomeriggio

Moderatore: Pierpaolo Mastroiacovo

13:10 – 13:30: Problemi psichiatrici e funzionamento intellettuale nella SVCF durante lo sviluppo; *Doron Gothelf, Bronwyn Glaser, Tamar Green, Stephan Eliez, Israel and Svizzera*

13:30 – 13:45: Frequenza di psicopatologie in bambini e adolescenti in Italia con SVCF: risultati preliminari del follow up di uno studio multicentrico; *Paola Atzori, Mariarosaria Scordo, Simona Teatini, Alida Varrella, Raffaella Turchi, Claudia Simonetti, Alessandro Zuddas, Italia*

13:45 – 14:00: Aspetti e caratteristiche psicopatologiche in bambini e adolescenti italiani con SVCF: risultati preliminari di uno studio del follow up su 15 pazienti; *M.R. Scordo, S. Teatini, A. Melani, R. Turchi, A. Varrella, C. Simonetti, Italia*

14:00 – 14:15: Monitoraggio della realtà e capacità nel riconoscimento delle emozioni nella SVCF; *Maude Schneider, Martin Debbané, Bronwyn Glaser, Catherine Pasca, Martial Van der Linden, Stephan Eliez, Svizzera*

14:15 – 14:30: Studio longitudinale delle difficoltà neuropsicologiche, comportamentali e psichiatriche nella SVCF; *Martin Debbané, Bronwyn Glaser, Marie Schaer, Stephan Eliez, Svizzera*

14:30 – 14:40: Profilo cognitivo di 20 bambini con SVCF; *M Bua, S Meduri, P Facchin, L Visonà, E Rizzardi, S Marinello, EM Ruga, Italia*

14:40 – 14:50: Domande e discussione

14:50 – 15:05: Pausa Caffè

Moderatore: Paolo Rossi

15:05 – 15:20: Come i genitori percepiscono i comportamenti del sonno e i disturbi del sonno di bambini con SVCF e dei loro fratelli; *Ann Swillen, Katrijn Van Aken, Jean-Pierre Fryns, Walter Hellinkcx, Koen Devriendt, Belgio*

15:20 – 15:35: Densità delle ossa e metabolismo in soggetti con SVCF; *Elisabetta Lapi, Stefano Stagi, Eleonora*

Gambineri, Roberto Salti, Maurizio Genuardi, Gloria Colarusso, Camilla Conti, Maurizio de Martino, Chiara Azzari, Italia

15:35 – 15:50: Funzione della tiroide e morfologia in soggetti con SVCF; *Stefano Stagi, Elisabetta Lapi, Eleonora Gambineri, Maurizio Genuardi, Gloria Colarusso, Camilla Conti, Rita Jenuso, Chiara Azzari, Maurizio de Martino, Italia*

15:50 – 16:05: Crescita e alimentazione; *M.Cristina Digilio, Rossella Capolino, Paola Cambiaso, Luigi Tarani, Marco Cappa, Bruno Marino, Bruno Dallapiccola, Italia*

16:05 – 16:20: Alimentazione e crescita nella SVCF: una curva di crescita per la SVCF e sue implicazioni genomiche; *Robert J. Shprintzen, USA*

16:20 – 16:30: SVCF e cardiopatie congenite: durata delle ospedalizzazioni post-operatorie; *E.M. Ruga, S. Meduri, R. Biffanti, G. Stellin, E. Rizzardi, S. Marinello, O. Milanese, Italia*

16:30 – 16:45: Domande e discussione

Domenica mattina, 9 Luglio 2009

Moderatore: Philippe De Clercq (Francia)

9:00 – 9:30: Strategie educative per bambini con SVCF; *Donna Cutler-Landsman, USA*

9:30 – 9:50: Il Progetto Inclusione e Opportunità di Apprendimento (ILOP): una breve rassegna e un progetto di ricerca ; *Maurizio Gentile, Simona D'Alessio, Eva Pigliapoco, Ivan Sciapeconi, Enrico Sitta, Francesco Pisanu, Italia*

9:50- 10:05: Risultati preliminari sulla tempistica del movimento di bambini delle scuole elementari con SVCF e di un gruppo di controllo della stessa età e stesso QI; *Katrijn Van Aken, Bouwien Smits-Engelsman, Ann Swillen, Belgio*

10:05- 10:20: Profilo neuropsicologico di un gruppo di bambini con SVCF in Italia ; *Deny Menghini, Francesca Addona, Manuela Mantovan, Stefano Vicari, Italia*

10:20 – 10:30: Domande e discussioni

10:30 – 10:45: Pausa Caffè

Moderatore: Stephan Eliez (Svizzera)

10:45 – 10:50: La Legislazione del Texas e la sindrome VCF; *Dianne Altuna, M.S., U.S.A.*

10:50- 11:05: Avvicinare la ricerca di nuovi farmaci e di sviluppo per il pubblico in Europa e Stati Uniti d'America; *Stuart Kushner, M.D., USA*

11:05- 11:15: Domande e discussione

11:15–12:00 Presentazione delle Associazioni Internazionali e relazioni dei Direttori Regionali di Europa, Stati Uniti e Canada, Messico e America Latina, Israele e Medio Oriente, Australia ed Estremo Pacifico.

12:00 Saluti dagli Organizzatori e Aggiornamento

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