La Sindrome di Williams Beuren (SW), descritta per la prima volta dal Dott. J.C.P. Williams nel 1961 in Nuova Zelanda, è una malattia genetica rara, non degenerativa, con incidenza di1/10.000 nascite. Essa consiste in un disordine neurocomportamentale congenito, dovuto alla delezione del gene dell'elastina nel cromosoma 7. Non è una malattia ereditaria, non è causata da fattori medici, ambientali o psicosociali ma piuttosto rappresenta "un caso". La SW interessa diverse aree dello sviluppo tra cui quella cognitiva, del linguaggio e psicomotoria.

L'Associazione Italiana Sindrome di Williams (AISW) nasce nel giugno del 1996 grazie alla volontà di un gruppo di genitori col proposito di realizzare senza fini di lucro la soluzione dei problemi medici, riabilitativi, educativi sociali e legali delle persone affette da SW e delle loro famiglie. Grazie all'attività del comitato scientifico e direttivo, all'apporto di professionisti altamente qualificati, e al prezioso lavoro dei volontari, si sono fatti fondamentali passi avanti.

Dal 2004 l'AISW collabora attivamente con le altre associazioni europee grazie alla fondazione della FEWS, federazione che le riunisce e le coordina.

The Williams Beuren Syndrome (SW), described for the first time by Dr. J.C.P. Williams in 1961 in New Zealand, is a rare genetic disease, not degenerative, with an incidence of 1/10,000 births. It consists of a neurobehavioral congenital disorder, due to the deletion of the elastin gene on chromosome 7. It is not an inherited disease, it is not caused by medical, environmental or psychosocial factors but it is rather "by case". The SW affects several areas of development including the cognitive, language and psychomotor areas.

The Italian Association for Williams Syndrome (AISW) was founded in June 1996 by the will of a group of parents with a desire to create a non-profit solution to solve medical, rehabilitative, educational, social and legal problems of the people with SW and their families. Thanks to the activities of the Scientific Committee and Board, the contribution of highly qualified professionals, and the valuable work of volunteers, fundamental progress has been made.

Since 2004 the AISW works closely with other European associations with the founding of FEWS, the federation that unites and coordinates them.

AISW
wishes to thank

Bambino Gesù
OSPEDALE PEDIATRICO

CIRCOLO S. PIETRO

the NANDO PERETTI foundation

FEWS
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ASSOCIAZIONE ITALIANA
SINDROME DI WILLIAMS

Harmonize Williams Syndrome in Europe

Rome · 17th October 2015

Pontificia Università Lateranense
Aula Magna
Piazza San Giovanni Laterano, 4
00120 Rome

WWW.HWSIE.AISW.IT

08.30/09.00 **REGISTRATION**

09.00/09.30 **OPENING CEREMONY**

Leopoldo Torlonia, President of the Italian Williams Syndrome Association

Mariella Enoc, President of the Bambino Gesù Pediatric Hospital

Giuseppe Novelli, Rector of Tor Vergata University

Camelia Lazar Livieratou, President of FEWS

09.30/10.30 **CLINICAL ASPECTS & PATIENTS**

Session Chair: **Bruno Dallapiccola,** Scientific Director of the Bambino Gesù Pediatric Hospital, Rome, Italy

Maria Cristina Digilio, MD, Chief of Medical Genetics Unit, Bambino Gesù Pediatric Hospital, Rome, Italy

Rare clinical features in Williams Syndrome.

Laura Mazzanti, MD, Pediatric Endocrinology and Rare Disease Unit, S.Orsola Malpighi University Hospital, University of Bologna, Bologna, Italy

Transition to Young Adulthood in Williams Syndrome.

Angelo Selicorni, Pediatrician and Medical Geneticist, Director of Pediatric Clinic Genetics Unit, MBBM Foundation, Monza, Italy

Old and new informations regarding the Williams Syndrome's natural history.

10.30/11.00 **COFFEE BREAK**

11.00/12.00 **GENETICS**

Session Chair: **Marco Tartaglia,** Research Division "Genetic Disorders and Rare Diseases", Bambino Gesù Pediatric Hospital, Rome, Italy

Luis A. Pérez Jurado, Professor of Genetics, Universitat Pompeu Fabra, Hospital del Mar Research Institute (IMIM) and CIBERER, Barcelona, Spain

Identifying the main genes and modifiers of the WBS phenotype through the characterization of patients and mouse models.

Giuseppe Merla, Medical Genetics Units IRCCS Casa Sollievo della Sofferenza Hospital, San Giovanni Rotondo, Italy

Understanding the Williams Beuren Syndrome by cell modeling and high throughput techniques.

Pierre Luc Germain, Laboratory of Stem Cell Epigenetics, European Institute of Oncology, Milan, Italy.

Dissecting the interplay of genetic and epigenetic mechanisms in Williams Syndrome: insights from cell reprogramming-based disease models.

12.00/12.45 **QUESTION TIME**

12.45/13.00 **POSTER PRESENTATION**

13.00/14.00 **BUFFET LUNCH**

14.00/15.00 **NEUROPSYCHOLOGICAL ASPECTS**

Session Chair: **Andrea Bartuli,** Head of Diagnosis, Treatment and Research in Patients affected by Rare and Genetic Diseases, Bambino Gesù Pediatric Hospital, Rome, Italy

Stefano Vicari, MD, Head of Child Neuropsychiatry Unit, Department of Neuroscience, I.R.C.C.S. Bambino Gesù Pediatric Hospital, Rome, Italy

Neurocognitive and behavioral aspects (infants and adults).

Annette Karmiloff – Smith, Professorial Research Fellow at Birkbeck's Centre for Brain and Cognitive Development, University of London, UK

Studying cognitive development in babies with Williams Syndrome compared to other neurodevelopmental disorders.

Chiara Gagliardi, MD, IRCCS Eugenio Medea, Bosisio Parini, Italy

Neurobiological basis of socio-emotional behaviour in Williams Syndrome.

15.00/16.00 **MEETING THE EUROPEAN WILLIAMS ASSOCIATIONS**

16.00/16.45 **OPEN TALKS**

Domenica Taruscio, CNR Director - Istituto Superiore Sanità

Danila Baldessari, Grant Manager - Fondazione Telethon

President, UNIAMO FIMR Onlus

16.45/17.30 **QUESTION TIME**

17.30/18.00 **CONCLUSION**

The conference language is English.

Simultaneous translation into Italian will be provided.