

# Final Program

## Sunday, April 17

3.00 p.m. – 5.30 p.m.	AISS meets Italian Scientific Committee
5.00 p.m.	Registration and Poster Set-up
6.30 p.m. – 9.00 p.m.	Opening - Palazzo della Gran Guardia

### **AISS meets Italian Scientific Committee**

*Sunday 17, 15:00-17:30*  
*Palazzo della Gran Guardia – Verona*

**Moderatori** : *Cesare Danesino e Sandra Perobelli*

15:00	<i>M.Cipolli</i>	Introduzione e Aspetti Clinici
15:15	<i>S.Cesaro</i>	Aspetti Ematologici
15:30	<i>F.Lavini</i>	Aspetti Ortopedici
15:50	<i>M.Daldoss</i>	Aspetti Nutrizionali
16:00	<i>S.Perobelli</i>	Aspetti Psicologici
16:10	<i>Compri</i>	Aspetti Odontoiatrici
16:30	<i>C.Danesino</i>	Aspetti Genetici
16:45	<i>M.Cipolli</i>	News di Ricerca
17:00		Discussione

\*Representatives of all International Association/Foundation are welcome

\*Simultaneous English translation will be available

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## Monday, April 18

8.15 a.m.	Poster Set-up
8.30 - 8.45 a.m.	Opening and Welcome
8:45 a.m. – 10.30 a.m.	<b>Clinical Aspects 1</b> <i>Moderator : J.M. Liu (US)</i>
8:45 a.m.	Clinical Phenotype and Overview <i>M. Cipolli (IT)</i>
9:00 a.m.	Hemathological Manifestation <i>J.M. Liu (US)</i>
9:30 a.m.	Skeletal Abnormalities <i>E. Samaila – F. Lavini (IT)</i>
10:00 a.m.	Growth and Nutritional Status <i>M. Daldoss (IT)</i> (Abs. 33)
10:10 a.m.	Bone Defects, Cellular or Systemic Impairment? <i>A. Frattini (IT)</i> (Abs. 02)
10:20 p.m.	Anaemia and Malabsorption Presenting in a Male Infant at Two Months of Age <i>E. Bakkeheim (NO)</i> (Abs. 05)
<i>10:30 – 11:00 a.m.</i>	<i>Coffee break</i>
11:00 a.m.- 12.45 p.m.	<b>Clinical Aspects 2</b> <i>Moderator: B.P. Alter (US)</i>
11:00 a.m.	An Outlook on Adult life in SDS <i>S. Perobelli</i>
11:15 a.m.	Fertility and Pregnancy in Inherited Bone Marrow Failure Syndromes <i>B.P. Alter (US)</i>
11:35 a.m.	Long Term Survival Probability in SDS Italian Patients <i>G. Tridello (IT)</i>
11:50 a.m.	Validated programs to support cognitive development <i>E. Kerr (CA)</i>
12:10 p.m.	Bone Marrow Characteristics in SDS and Hematological Complications <i>B. Zlateska (CA)</i> (Abs. 23)
12:20 p.m.	Cognitive Interactional Assessment in Children and Teenagers with SDS <i>M. Batt (FR)</i> (Abs. 04)
12:30 p.m.	Misdiagnosis and/or Mistreatment of Children with SDS <i>E. Tsangaris (CA)</i> (Abs. 08)

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12.45 p.m. - 1.45 p.m.	Lunch
1.45 p.m. - 2.45 p.m.	Poster View
2.45 p.m. - 3.30 p.m.	<b>Lecture</b> <i>S. Desrivieres (UK) :</i> "Genetic Markers Of Brain Alterations And Neurocognitive Abilities In Health And Disease: Promises And Challenges Of Neuroimaging Genomics"
3:30 p.m. - 4:00 p.m.	Coffee break
4:00 p.m. - 5:15 p.m.	<b>Roundtable on SDS Registry</b> <i>Moderators: P. Durie (CA) and B.M. Assael (IT)</i> <i>Speakers: J. Donadieu (FRA), A. Shimamura (USA), Y. Dror (CA) and M. Cipolli (IT)</i>
5:15 p.m. - 6:00 p.m.	<b>Selected abstracts</b> <i>moderator: Y. Dror (CA)</i>
5:15 p.m.	Uncommon Acquired Chromosome Anomalies in SDS <i>E. De Paoli (IT) (Abs. 16)</i>
5:30 p.m.	The North American SDS Registry: Genetically Undefined SDS <i>K.C. Myers (US) (Abs. 07)</i>
5:45 p.m.	Distribution of Genetic Variants rs4906844 and rs11633924 in a Group of Patients with SDS and Correlation with Cognitive Impairment <i>L. Nacci (IT) (Abs. 36)</i>

# Final Program

## Tuesday, April 19

8:30 a.m. -10:30 a.m.	<b>Basics Science and Disease Models 1</b> moderator: J. Rommens (CA)
8:30 a.m.	Modeling Bone Marrow Failure and MDS in SDS using Induced Pluripotent Stem Cells <i>A. Shimamura (US)</i>
9:00 a.m.	New Insights into the SDS-related Haematological Disorder: Hyper-activation of mTOR and STAT3 in Leukocytes <i>V. Bezzeri (IT)</i>
9:30 a.m.	SBDS Protein in Rybosome Assembly <i>A. Warren (UK)</i>
10:00 a.m.	Deletion of Mitochondrial Porin Alleviates Stress Sensibility in the Yeast Model of SDS <i>A.N. Jensen (TH) (Abs. 19)</i>
10:15 a.m.	Is the Loss of Interaction between the Proteins EFL1 and SBDS of SDS? <i>N. Sanchez-Puig (MX) (Abs. 18)</i>
10:30 a.m.	Deficiency of Elongation factor like GTPase 1 (Efl1) mimics aspects of Shwachman-Diamond Syndrome in the mouse <i>A. Hoslin (UK) (Abs. 39)</i>
10:45 a.m.-11:15 a.m.	<i>Coffee break</i>
11:15 a.m. - 12:45p.m.	<b>Basic Science and Disease Models 2</b> moderator: A. Warren (UK)
11:15 a.m.	Responses of Different Organs to Loss of SBDS as Revealed by Mouse Models <i>J. Rommens (CA)</i>
11:45 a.m.	A mammalian model of neutropenia in SDS <i>H.G.P. Raaijmakers (NL)</i>
12:15 p.m.	A Replication Stress-Induced Checkpoint Arrests Development in a Drosophila Model Of SDS <i>S. Tan (UK) (Abs. 11)</i>
12:30 p.m.	SBDS Knockout in Zebrafish Results in Neutropenia, Pancreas and Liver Atrophy, Diminished Growth and Reduced Viability <i>U. Oyarbide (US) (Abs. 12)</i>
12.45 p.m. - 2.00 p.m.	<i>Lunch</i>
2.00 p.m. - 3.00 p.m.	poster view

# Final Program

## Tuesday, April 19

3:00 p.m. - 4:15 p.m.	<b>Therapies</b> <i>moderator: B.M. Assael (IT)</i>
3:00 p.m.	Hematopoietic stem cell transplantation in Shwachman-Diamond Disease: an update <i>S. Cesaro (IT)</i>
3:15 p.m.	SDS hematopoietic system and its relevance to bone marrow failure and leukemia <i>Y. Dror (CA)</i>
3:30 p.m.	SBDS and Basic Defect Therapy <i>M. Cipolli (IT)</i>
3:45 p.m.	Outcome of Hematopoietic Stem Cell Transplantation in Patients with SDS with or without Secondary MDS/AML: an Analysis of the German Shwachman Diamond Cohort by the SCNIR-Europe Mellor <i>S. Mellor-Heineke (DE) (Abs. 09)</i>
4:00 p.m.	Multidisciplinary Care and Stem Cell Transplantation for SDNS: the Ste-Justine experience <i>Y. Pastore (CA) (Abs. 10)</i>
4:15 p.m. - 4.45 p.m.	<i>Coffee Break</i>
4:45 p.m. – 5:30 p.m.	<b>Selected abstracts</b> <i>moderator: S. Cesaro (IT)</i>
4:45 p.m.	Premature Senescence in Hematopoietic Stem and Progenitor Cells in Ribosomopathy-Associated Bone Marrow Failure Syndromes <i>J.M. Liu (US) (Abs. 15)</i>
5:00 p.m.	SDS Murine Model Organs Exhibit Reduction of 80S Ribosomes and Heavy Polysomes with Altered Transcript Binding <i>J. Rommens (CA) (Abs. 14)</i>
5:15 p.m.	Expression Studies in Bone Marrow of Patients with SDS Carrying a Chromosome 20 long Arms Interstitial Deletion <i>R. Valli (IT) (Abs. 13)</i>

# Final Program

## Wednesday, April 20

8:30 a.m. -11:00 a.m.	<b>Genetics</b> <i>moderator: C. Danesino (IT)</i>
8:30 a.m.	Cytogenetic Monitoring of SDS <i>F. Pasquali (IT)</i>
9:00 a.m.	Ribosomopathies and the paradox of cellular hypo- to hyperproliferation <i>K. De Keersmaecker (BE)</i>
9:30 a.m.	Preliminary results of exome sequencing in SDS <i>C. Danesino (IT)</i>
10:00 a.m.	Genetic Analysis of the SBDS Gene among Non-Italian Patients <i>E. Nicolis (IT) (Abs. 20)</i>
10:15 a.m.	Genetic Variation in the SDS Gene, SBDS <i>J. Rommens (CA) (Abs. 01)</i>
10:30 a.m.	Small-Angle-X Ray-Scattering Studies of the Low-Resolution Structure of the Ribosomal GTPase EFL1, the SBDS Protein and Their Complex <i>D. Siliqi (IT) (Abs. 21)</i>
10:45 a.m.	Radiosensitivity and DNA Damage Repair in Lymphoblastoid Cell Lines Derived from SDS patients <i>J. Morini (IT) (Abs. 24)</i>
11:00 a.m.- 11.30 a.m.	<i>Coffee Break</i>
11:30 a.m.-12:30 p.m.	<b>Closing Remarks:</b> <ul style="list-style-type: none"> <li>- <b>Families point-of-view</b></li> <li>- <b>Summary and perspectives</b></li> <li>- <b>Next meeting announcement</b></li> </ul>