

# KLEEFSTRA ITALIA CONFERENCE

*Saturday, 17 September 2022*

*on the occasion of the*

***World Day***

***of awareness of Kleefstra Syndrome***

*the Italian KS Association*

*organizes the second Conference*

*in the Metropolitan City of Naples,*

*precisely in the Municipality of Pozzuoli*

*at **TIGEM** - Telethon Institute of Medical Genetics*

*with the patronage of Fondazione Telethon.*

F O N D A Z I O N E



SINDROME  
DI **KLEEFSTRA**  
ITALIA

## ***The Conference aims to:***

- ★ *Bringing together families with KS children and young people, who are currently a total of 58 in Italy*
- ★ *Acquiring information and data regarding Kleefstra Syndrome from Geneticists, Neuropsychiatrists and Professionals.*
- ★ *Sharing information, experiences and research progress*
- ★ *Collecting, directly from the therapists who follow the children, information on rehabilitation treatments*

# Organization

**Friday 16 September:** Welcome and dinner in Agave Hotel

**Saturday 17 September:** Conference, takeaway lunch at the Tigem and dinner in Agave Hotel.

**Sunday 18 September:** Moments of sharing, lunch, departure

All participants will stay at:

✓ **Agave Hotel**

SS 7/IV Domitiana

Km 53+390

80078 Pozzuoli, Italia

tel.: +39 081 524 9961

website: [www.agavehotel.it](http://www.agavehotel.it)

email: [info@agavehotel.it](mailto:info@agavehotel.it)

✓ **Hotel Villa Luisa**

Via Tripergola 50 - 80078 Pozzuoli (NA)

+ 39081 8042870

+ 39081 8042852

email: [info@villaluisaresort.it](mailto:info@villaluisaresort.it)

The Conference will take place in the Hall at the Tigem - Telethon Institute of Medical Genetics, in Pozzuoli.

For reservations and information regarding rooms, meals and transport to and from airports (Naples, is the nearest airport), to and from the train stations (the nearest station is Naples) please contact the Resort.

## **COSTS TO BE BORNE BY HOUSEHOLDS:**

Food and lodging at the Resort  
Various transports to arrive and leave.

# Entertainment for children and teenagers



*Children can play outdoors. There will be an entertainment with animators of the Melofiocolo, Educational Farm.*

*Each family can bring board games and/or small musical instruments of interest to their child.*

## **PLEASE NOTE -IMPORTANT**

*There are no professional educators.  
Family members will have to supervise  
their **children.***

# CONFERENCE PROGRAMME

**Friday September 16, 2022**

Arrival day: informal meeting at the hotel, dinner

**Saturday, September 17, 2022**

From 8:15 am **IMPORTANT:**

***Registration, delivery of headsets with ID deposit. Each user is responsible for the headsets while remaining at his own expense for any damage.***

Moderator: Prof. Nicola Brunetti-Pierri

**9.10 -9.15** *"Introduction to Tigem"*

**Dr. Valentina Bouché**

PhD – Telethon Institute of Genetics and Medicine Scientific Officer

**09:15 – 09:30 Dr. Mariella Priano and Dr. Orsola Rizzardi:**

*"Greetings and Introduction"* – Kleefstra Syndrome Association Italy

**09:30 – 10:00 Dr. Tjitske Kleefstra**

*"History and genetic actuality of Kleefstra syndrome"*

**10:00 – 10:20 Arianne Bouman**

*"Studies and development of the syndrome"*

**10:20 – 10:50 Dr. Karlijn Vermeulen** in telematic connection

*"Psychiatric aspects of Kleefstra syndrome"*

**10:50 – 11:10 Prof. Valentina Massa**

*"Registry, biobank and experimental models for the study of Kleefstra syndrome: the Italian project"*

**Dr. Camilla Meossi**

*Answers questions about data collection and registry creation*

**11:10 – 11:30:** coffee break

**11:30 – 11:50 Prof. Angelo Selicorni** in telematic connection  
*"Definition of paediatric care problems for children with Kleefstra syndrome – Guidelines for follow-up"*

**11:50 – 12:10 Prof. Nicola Brunetti-Pierri**  
*"Animal models for the study of genetic diseases"*

**12:10 – 12:30 Prof. Giuseppe Merla**  
*"Genetics and etiology of chromatinopathies"*

**12:30 – 12:50 Dr. Alberto Danieli**  
*"The management of epilepsy in rare neurodevelopmental diseases"*

**13:00 - 14:30 Lunch**

**14:30 – 14:50 Dr. Ettore Caterino**  
*"Sleep troubles in KS. Italian evaluation protocol and proposals for the guidelines"*

**14:50 – 15:10 Dr. Rosa Siliotto**  
*"Speech therapy interventions in children with Kleefstra Syndrome"*

**15:10 – 15:30 Dr. Antonio Fimiani**  
*"The role of epigenetics in genetic disease"*

**15:30 – 15:50 Dr. Martina Castaldo**  
*"Potential of assisted animal therapies with a horse"*

**15:50 – 16:30 Pause**

**16:30 – 16:50 Luca Trapanese** in telematic connection  
*"After us: a feasible reality" in telematic connection*

**16:50 – 18:00** *"The experience of family and siblings"*

**18:00 – 19:00 Discussion/questions&answers**

**IMPORTANT:**  
**When finished, please return the audio material**

## ***Participants:***

- **Prof. Tjitske Kleefstra** MD PhD, Clinical Geneticist, Radboud University
- **Ariane Bouman** M.D., PhD student Department of Genetics Radboud
- **Dr. Karlijn Vermeulen** Child Psychiatrist, the Netherlands
- **Prof. Valentina Massa**, Associate Professor of Applied Biology – University of Milan
- **Dr. Camilla Meossi** Doctor specializing in Medical Genetics University of Milan
- **Prof. Angelo Selicorni** MD Director UOC, Pediatrician and Geneticist Ospedale Sant'Anna (Como)
- **Prof. Nicola Brunetti-Pierri**, MED, Tigem Pozzuoli e Federico II University of Naples, Italy
- **Prof. Giuseppe Merla** - PhD University of Naples Federico II – IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo
- **Dr. Alberto Danieli**, Neuropsychiatrist at IRCCS Eugenio Medea, Institute for Rare Diseases Research and Rehabilitation - Conegliano (TV)
- **Dr. Ettore Caterino** Child Neuropsychiatrist Psychotherapist – Head of the Autism Clinical Network ASL Sudest Toscana
- **Dr. Rosa Siliotto** Doctor in speech therapy and neurotherapist qualified to exercise myofunctional therapy, Verona and Brescia
- **Dr. Antonio Fimiani** Specialist in Posturology, Physiatry, Dermatology, Orthopedics
- **Dr. Martina Castaldo** psychologist, specialized in assisted interventions

## **Sunday 18 september 2022**

Breakfast

10:00 Relax by the swimming pool

13:00 Lunch

***Good by... Arrivederci!!!!***

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**ASSOCIAZIONE ITALIANA  
SINDROME DI KLEEFSTRA – APS**

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