





VIII international congress

CORNELIA DE LANGE SYNDROME

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Raríssimas would like to invite you all to participate on this important VIII International Cornelia de Lange Syndrome Congress.

As a mother, I would like to personally welcome you. A glad welcome filled with the desires, hopes and dreams that we all certainly share.

As many of you, I had CDLS son – Marco.

I was 20 and had been married for just over a year when I got pregnant. At the time, there were few means of diagnosis but I knew something was wrong.... But, when he was born, I looked at his little face and I did not see anything different. For me, it was the most beautiful baby in the world!

Later, when the doctor told me he had CDLS, my moments of anguish began. No one could answer my questions, so my fight was personal! I did not traced plans for the future because I did not know what to expect. So I decided I had to enjoy every minute I had with my son...

Today, thankfully we have new drugs, new means of diagnosis and new perspectives about this rare disease. So we invite you all to discuss how to achieve our children's potential, regardless their disease. Side by side, medical, scientists and parents will fight in order to bring a brighter future to all the marcos of all over the world.

We are waiting for you.

Prepare yourself to be challenged, excited and inspired!

See you all soon!



Cornelia de Lange

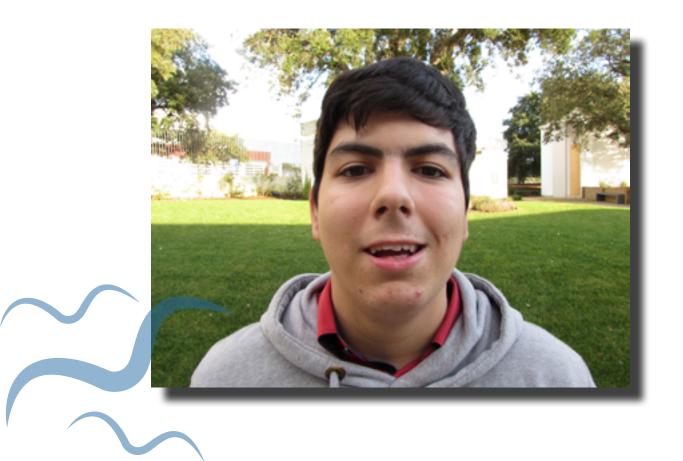
Cornelia de Lange syndrome (CdLS) is a multisystem disorder with variable expression marked by a characteristic facial dysmorphism, variable degrees of intellectual deficit, severe growth retardation beginning before birth (2nd trimester), abnormal hands and feet (oligodactyly, or sometimes an even more severe amputation, and constant brachymetacarpia of the first metacarpus), and various other malformations (heart, kidney etc.).

The prevalence in the general population is estimated at between 1/62,500 and 1/45,000.

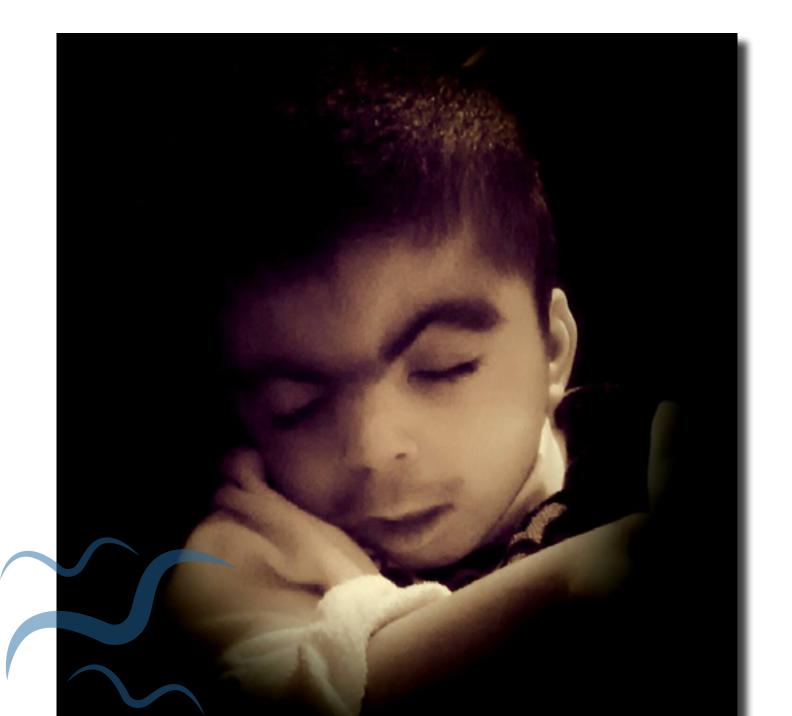
Distinctive facial features include: well-defined curved and confluent eyebrows, long eyelashes, anteverted nares, micrognathia, and downturned corners of the mouth with a thin upper lip. Feeding problems and failure to thrive are frequently complicated by gastroesophageal reflux. Clinical progression leads to psychomotor retardation, language acquisition difficulties and, sometimes, behavioral disorders in the autistic spectrum. Hearing loss should be evaluated and monitored.

Causative mutations in three genes involved in chromosomal cohesion (cohesin complex) have been identified. The NIPBL gene is mutated in approximately 50% of patients and is the major gene involved in the syndrome. Mutations associated with milder forms of the disease have recently been described in the SMC1L1 gene (also called SMC1A; Xp11.22-p11.21), associated with an X-linked form of CdLS, and in the SMC3 gene (10q25).

Prenatal diagnosis may be suspected after a prenatal ultrasound examination revealing intrauterine growth retardation and limb defects. In families with parental transmission of the disease, and in which a mutation has been identified, prenatal diagnosis by analysis of fetal DNA can be proposed, even to parents who do not appear to carry the mutation because of the risk of germinal mosaicism.



Clinical progression leads to psychomotor retardation, language acquisition difficulties and, sometimes, behavioral disorders in the autistic spectrum. Hearing loss should be evaluated and monitored



CdLS World Federation

CdlS World has been founded in 2001 in occasion of the first International CdLS Conference and counts many members (Argentina, Australia, Canada, Denmark, France, Germany, Italy, Japan, Netherlands, Poland, Portugal, Spain, UK, USA) and is maintaining contact with single families in more than 45 countries. We welcome groups that would like to join the federation.

From all over the world the Experts who know most about CdLS and a specific topic have formed a international Scientific Advise Committee (SAC). We make sure the SAC is informed about needs of the families and caretakers, so they can publish relevant news, present on conferences and answer questions in our Ask the Expert service.

Raríssimas

Raríssimas was founded in 2002 by a group of mothers of children suffering from a rare disease. It was registered as an NGO in 2003. Its headquarters are in Lisbon, but there are two other Delegations (Oporto and Azores). Raríssimas has 2 Rehabilitation Centres and a Helpline - Linha Rara, distinguished .for the 2nd year at a European level. Raríssimas is a founding member of FEDRA – Portuguese Federation of Rare Diseases Associations, a member of Eurordis and Cornelia de Lange Syndrom Foundation.

One of the Association main achievement is the project "Casa dos Marcos", a sheltering home for adults and/or young/adults, bearers of rare diseases, lacking in support and in intellectual or leisure activities. The Resources Centre, located in Moita, near Lisbon, is also a home for sick persons suffering rare pathologies after the death of their parents. It is has been conceived as a space where people affected by a rare disease can develop the skills that will allow them to integrate into society and live lives that are as normal possible. The Casa dos Marcos also serves as a residence for those that require intensive care.









Scientific Symposium Catholic University - Lisbon CDLS -Rare diseases paradigm

8 a.m.	// Registration
8:45 a.m.	// Welcoming and Opening Ceremony • Maria da Glória Garcia Rector of Catholic University of Portugal
	 Alexandre Castro Caldas Health and Science Institute of Catholic University of Portugal
	• Paula Brito e Costa CDLS World Federation Chair, Raríssimas President
	• Portuguese Republic President- To Be Confirmed (TBC)
	• José Manuel Silva President of the Portuguese Medical Association
	• Germano Couto President of the Portuguese Nurses Association
	• Luís Portela Health Cluster
9:15 a.m.	// Health research and innovation framework in the European context • Panel Moderator: Emília Monteiro
	Portuguese Clinical Research Infrastructure Network – PtCRIN
	• Maria Graça Carvalho European Commission DG Research and Innovation - TBC
	• (TBC) European Commission DG Health and Food Safety



9:45 a.m.	// Frameworks of rare diseases in the European and National context
11:15 a.m.	// Coffee Break
11:30 a.m.	// Framework for orphan medicinal product in the European context
1 p.m.	// Lunch

2:30 p.m.	// CDLS World – Our cases
3:15 p.m.	// Cornelia de Lange syndrome clinical course – Rare Disease Example (I)
4:15 p.m.	// Cornelia de Lange syndrome clinical course – Rare Disease Example (II) • José Pedro Vieira Neurological disorders • Peter Gillett Gastroesophageal reflux (TBC) • Alessandra Moretto Problems / complications after the surgery (TBC)









5:15 p.m.	// Coffee Break
5:30 p.m.	// Cornelia de Lange syndrome clinical course – Rare Disease Example (III) • (TBC) Psychomotor evaluation and Rehabilitation • Antonella Constantinou How to promote Communication (TBC) • Emanuele Basile Family Burden
6:30 p.m.	// Closing Ceremony Portuguese Republic President (TBC)



Accommodation of participants on the meeting

LISBON MARRIOTT HOTEL

AVENIDA DOS COMBATENTES, N.º 45 LISBOA

Phone number: +351 217 235 400

www.lisboamarriott.com

Enjoy the style and quality of a 4-star superior hotel with personalized service. Located in the new financial district, a short distance from Lisbon airport and the old town, our hotel gives guests easy access to the entire city. Exquisitely decorated and with functional rooms and suites we offer picture windows and a luxurious and unique comfort. High speed internet access in rooms and public areas, swimming pool in the garden and 24-hour gym are additional benefits to enhance your stay in Lisbon.

The Citrus Bar and Restaurant offers a Portuguese Atlantic influence kitchen with a fine selection of local and international wines. Connected to the garden terrace you can enjoy your meal while enjoying the warm sun of Lisbon. You will appreciate the 2,000 square meters of our congress center. Rooms and elegant and versatile spaces for meetings, events and exhibitions as well as a specialized and professional staff attentive to every detail. The congress center has 16 meeting rooms in a flexible area of 2000m2 and can accommodate large and small events of various kinds, from meetings and conferences to product presentations, exhibitions and ceremonies.

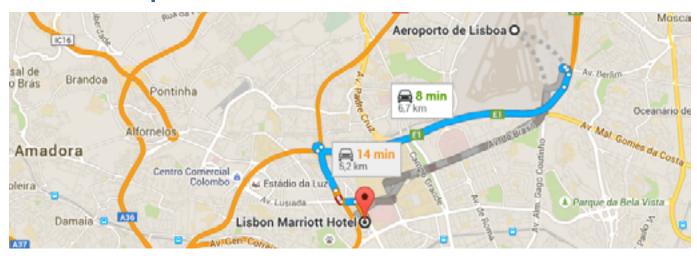








Lisbon airport -> Lisbon Marriott Hotel



Lisbon Marriott Hotel -> Catholic University





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Europear - 20% discount (please mention contract n.º40489416)

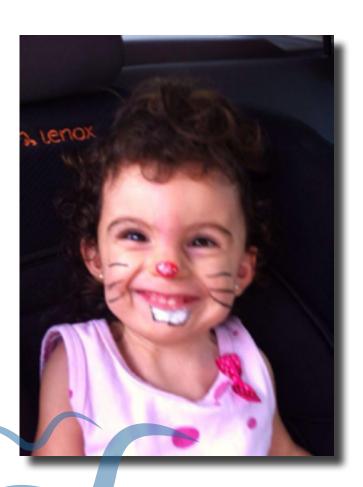
Reserves:

Tel.: 00351219407790

Email: reservas@europcar.com

Web: europcar.pt







Families Conference **Aqualuz Suite Tróia - Carvalhal**





11 a.m.	// Coffee Break
11:15 a.m.	// Genetics II • Prenatal diagnosis Frank Kaiser Germany Head of Section for Functional Genetics at the Institute for Human Genetics University of Lübeck • How genetics can help us - now and in the future Margarida Reis Lima Portugal Raríssimas SAC • Why clinical diagnosis is so important: follow-up's and emergency care protocols Angelo Selicorni Italy SAC • Discussion
1 p.m.	// Lunch
2:30 p.m.	// Global Challenges I
4 p.m.	// Coffee Break

4:15 p.m.	// Global Challenges II • Neuropsychiatric care José Pedro Vieira Portugal
	 Resistance to change and spectrum dependency among children with CdLS Bjørg S. Hoem Norway
	• The burden for a family Maria do Céu Seabra Portugal Emanuele Basile Italy
	• Discussion
6 p.m.	// Closing
8 p.m.	// Gala Dinner







9 a.m.	// CdLS Particular Clinical Issues
	Anesthesia and Cdls: risks and false fears Alessandra Moretto Italy
	 Medical problems to consider in a CdLS person: Presenting the clinical protocol Angelo Selicorni Italy SAC
	• Discussion
11 a.m.	// Coffee break
	PARALLEL SESSIONS
11:30 a.m.	room A // Classic – Paediatric Age on CdLS • Paediatric age on CdLS Luís Nunes Portugal Moderator • Medical follow-up Feliciano Ramos Spain • Rehabilitation approach Paola Ajmone Italy

11:30 a.m.

- Behaviour and communication questions Emanuele Basile | Italy
- School Challenges
 Mary Morse | United States of America
- Dental Issues
 Douglas Clemens
- Discussion

11:30 a.m.

room B

// Mild - Paediatric Age on CdLS

- Paediatric age on CdLS
 Salomé de Almeida | Portugal
 Moderator
- Medical follow-up Sylvia Huissman
- Dental Issues
 Douglas Clemens
- Rehabilitation approach Clara Almeida | Portugal
- Behaviour and communication questions
 Paul Mulder
- School Challenges
 Mary Morse | United States of America
- Discussion

rooms X to Z

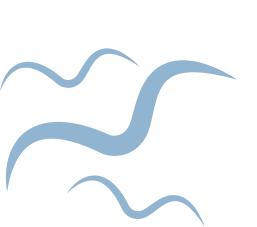
Clinical Consultations

1 p.m. // Lunch **PARALLEL SESSIONS** 2:30 p.m. room A // Classic - Young and adult age on CdLS Adult age on CdLS José Pedro Vieira | Portugal Moderator Medical follow-up Antonie Kline | United States of America SAC Sexuality Natalie Blagowidow | United States of America SAC After School Life – (whats now?) TBC | Italy Discussion room B 2:30 p.m. // Mild - Young and adult age on CdLS Adult age on CdLS Angelo Selicorni | Italy Moderator After School Life – (whats now?) Anna Contardi | Italy Medical follow-up Antonie Kline | United States of America SAC Sexuality Natalie Blagowidow | United States of America SAC Discussion rooms X to Z Clinical Consultations



4 p.m.	// Coffee Break
4:30 p.m.	// Answers for Future Life
6 p.m.	// Closing





Summer Camps for CdLS patients

Our Summer Camps have as mission to promote the full development and autonomy of children and young people with rare disease.

WE HAVE PROGRAMS THAT WILL MAKE YOUR SON'S DAY SHINE, ALL FOR FREE.

If you have another child (without CDLS) you can also enroll him in the summer camps. In this case, it will be charged 175 euros.

CAMP COORDINATORS **Andreia Bernardo e César Costa**CAMP ACCOUNTABLE **César Costa e Sofia Lopes**

PLEASE FILL HERE THE REGISTRATION FORM, IN ORDER TO REGISTER YOUR CDLS CHILD



Cornelia de Lange children

CDLS childcare – 11th and 12th free of charge – Aqualuz (* If needed on extra days, please contact **cdlsconference2015@rarissimas.pt**)

8:30 a.m.	// Welcoming
9 a.m.	// Welcoming presentation
10 a.m.	// Know each other time
12 a.m.	// Return to hotel
12:30 a.m.	// Epic meal time - Lunch*
13:30 a.m.	// Chill out session - After lunch
15:30 a.m.	// Beach - Relax (snacks included)
17:30 a.m.	// Sport activity

^{*} Lunch is not included in the program, it will be with their parents/careholders



8:30 a.m.	// Welcoming
9 a.m.	// Short welcoming presentation of the day
9:30 a.m.	// Beach
12 a.m.	// Epic meal time - Lunch*
13:30 a.m.	// Chill out session - After lunch
15:30 a.m.	// Epic pool time (snacks included)
17:30 a.m.	// Sayonara time

^{*} Lunch is not included in the program, it will be with their parents/careholders



Cornelia de Lange siblings children

8:30 a.m.	// Welcoming
9 a.m.	// Welcoming presentation
10 a.m.	// Know each other time
12 a.m.	// Return to hotel
12:30 a.m.	// Epic meal time - Lunch*
13:30 a.m.	// Chill out session - After lunch
15:30 a.m.	// Beach - Relax (snacks included)
17:30 a.m.	// Sport activity

^{*} Lunch is not included in the program, it will be with their parents/careholders



8:30 a.m.	// Welcoming	
9 a.m.	// Short welcoming presentation of the day	
9:30 a.m.	// Dolphin watching	
12 a.m.	// Epic meal time - Lunch*	
13:30 a.m.	// Chill out session - After lunch	
15:30 a.m.	// Epic pool time (snacks included)	
17:30 a.m.	// Sayonara time	

^{*} Lunch is not included in the program, it will be with their parents/careholders



Accommodation of participants on the meeting

AQUALUZ SUITE HOTEL APARTAMENTOS TRÓIA

TRÓIA - CARVALHAL GDL - TRÓIA 7570-789PORTUGAL

Phone number: +351 265 499 000

www.aqualuztroia.com

Modernity, comfort and functionality materialize into 365 apartments with contemporary decor in neutral tones and environments, combined with well-being at Wellness Centre (which has indoor and outdoor pools, sauna, jacuzzi, Turkish bath, spa services, fitness room) to provide unique stays.

At Aqualuz Tróia, while the little ones have fun at Troia kids, families can choose from a break in the Atrium Bar, a meal at Azimute Restaurant or one of many activities designed to discover the vast environmental and cultural heritage of the Peninsula, available throughout the year.

From the beaches to swimming pools, tennis courts, biking, walking, dolphin watching, golf, water sports and visits to Roman ruins, there are several options for different motivations.

Aqualuz Suite Hotel also offers a business center, six meeting rooms equipped with latest technologies and parking facilities, meeting the conditions in which working hours can be carried out efficiently and smoothly.













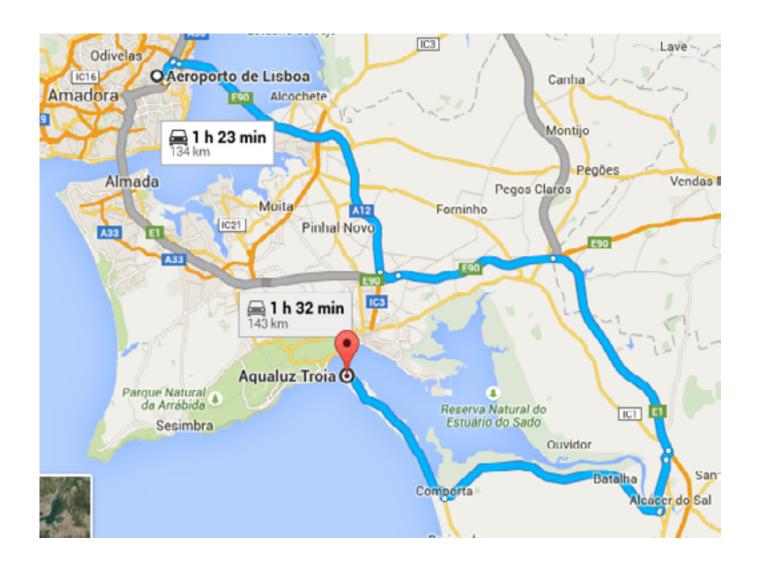








Lisbon airport -> Aqualuz Tróia





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Web: europcar.pt



What to visit

If you decide to spend a weekend, week or even a vacation in Tróia, know that there are numerous leisure activities that you can do. If you dont like the beach or the weather is not favorable, or if you prefer another type of leisure activities to help you relax in perfect harmony with the surrounding nature, Trojan offers immense alternatives to its visitors to get distracted and above all fun.

Read then some of the activities that you can accomplish if you decide to visit Troy.

Horse riding in Comporta

To perform a horseback ride may deal with the horses on the sand and investigate the possibilities within this type of activities. In addition to horseback riding on the beach, in the field or through the rice paddies and Sado estuary, know that here you can also make riding lessons if so desired.

The markings shall be made via email or telephone 48 hours in advance.

Email: info@cavalosnaareia.com Contact: (+351) 919 002 545

Dolphin watching

One of the most pleasant activities in Troia these are the boat trips for dolphin watching. Boarding with Vertigo Blue and leave this boat trip watching the bottlenose dolphins of the Sado and the natural beauty that makes up the estuary, Arrábida and the Troia peninsula. In summer you can even take a dip in the sea when the opportunity provides.

The matches are made from the port of Setúbal or Troia Marina and the tours usually last for three hours, so you can still spend your afternoon on the beach if you wish. Dolphins watching tours are held throughout the year.

Setúbal Edificio Marina Deck Health Beach Street 11D 2900-572 Setúbal - Portugal

Trojan Marina Tróia 7570-789 Carvalhal - Grândola Tel. 265 238 000 Tel. 916 982 907

Email: vertigemazul@mail.telepac.pt





Bird watching

For bird watching trip is by boat and the inside of the Sado estuary. This tour will allow you to observe the different species of birds that live there, accompanied by an expert guide that can clarify these interesting animals. The tours usually last for 3 hours and include an expert guide, binoculars and field guides. The period of emergence of these trips occurs from October to March.

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Trojan Marina Tróia 7570-789 Carvalhal - Grândola Tel. 265 238 000 Tel. 916 982 907 Email: vertigemazul@mail.telepac.pt







Catamaran tours

Throughout the year can still perform Catamaran tours off the coast of Tróia and Arrábida or inside of the Sado estuary. Catamaran sailboat on you can enjoy a peaceful environment and let yourself be carried away by the smell of the sea, the wind in the face and the many sensations and flavors resulting from this tour. During the summer you can even take place a stop so you can dive into the sea and cool off on hot days. These tours have a duration of 4 hours and include the boat rental and a buffet meal on board.

Setúbal Edificio Marina Deck Health Beach Street 11D 2900-572 Setúbal - Portugal

Trojan Marina Tróia 7570-789 Carvalhal - Grândola Tel. 265 238 000 Tel. 916 982 907

Email: vertigemazul@mail.telepac.pt



Meeting Chart

	Date	Price	Local
Scientific Simposium	9 th September	100€/Person	Catholic University
Families Conference	11 and 12 th September	255€/Person	Aqualuz Suite Tróia
Gala Dinner	11 th September	70€/Adults; 35€/Children	Aqualuz Suite Tróia
Summer Camps (brothers of CDLS Childs)	11 and 12 th September	175€/Children	Aqualuz Suite Tróia

Families' Conference subscribers have free registration fee at the Scientific Symposium.

Scientific Symposium includes:

Documentation and coffee-breaks.

Families' Conference includes:

Documentation, lunches (11th and 12th) and coffee-breaks, translation (dependent on a minimum number of requests).



General Information

Expected temperatures with highs of 26.° and lows of 22.°. These temperatures can, however, vary so it is recommended to check the same through the website of the Portuguese Institute of Sea and Atmosphere.

Link:

http://www.ipma.pt

Because of the temperatures this time of year, it is recommended cool clothes.

Essentials:

- Sunglasses
- Swimsuit / bikini
- Comfortable clothes for the tours
- Comfortable footwear for the tours
- 1 Formal Clothing
- Sun protector





CdLS Congress | SUPPORT

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VIII international congress

CORNELIA DE LANGE SYNDROME

9th-12th September 2015 **Lisboa | Tróia | Portugal**

REGISTRATION FORM HERE