Claudia Morozzi, M.D.

Department of Molecular Medicine,
"Sapienza" University of Rome, Italy

Extracorporeal Therapeutic Techniques Unit

Lipid Clinic and Atherosclerosis Prevention Centre

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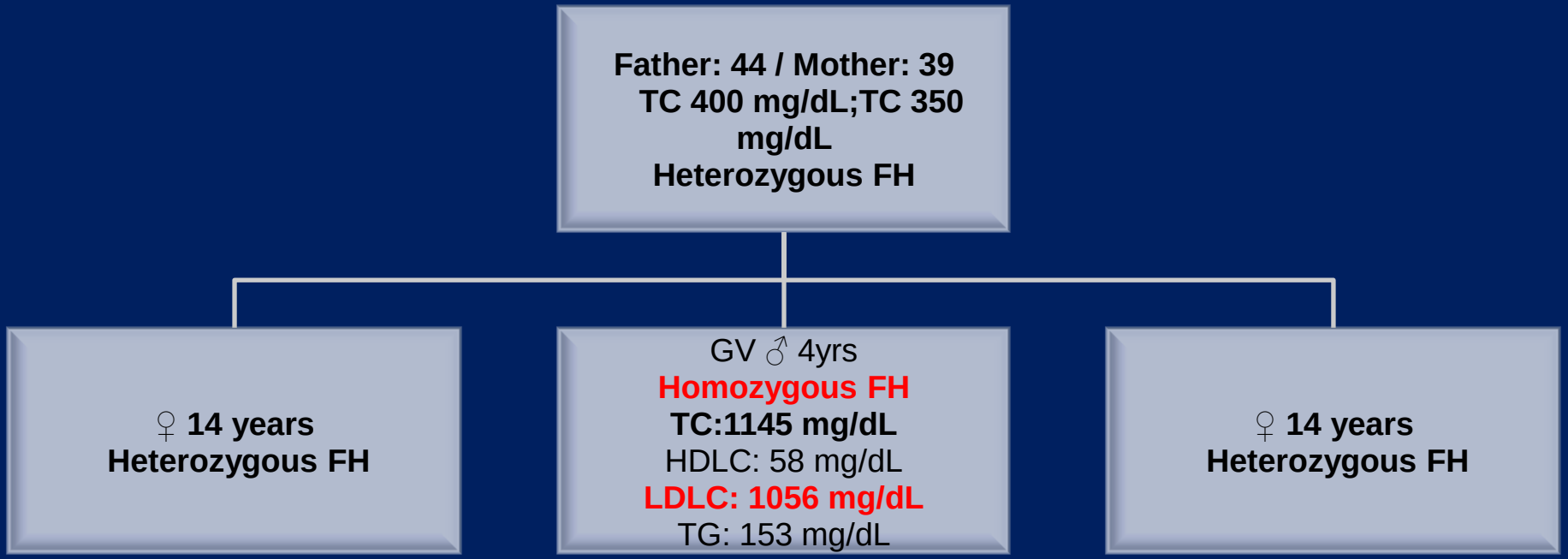
UMBERTO I
POLICLINICO DI ROMA

Homozygous Familial Hypercholesterolemia
GV ♂ : 4 years



SAPIENZA UNIVERSITÀ DI ROMA

UMBERTO I POLICLINICO DI ROMA

Homozygous Familial Hypercholesterolemia
GV ♂ : 4 years

- Diagnosed as to be HoFH when he was 1 year old but never submitted to any non invasive / invasive cardiovascular evaluation / imaging; Region: Campania

- After being evaluated at our centre, he was referred to cardiologists for CV assessment (imaging), per our protocol, before starting LA;

followed

The Pt reported symptoms (angina) at kindergarden and his parents called us. We asked them to bring immediately their son to local Hospital for intensive care. Unfortunately, the Doctors thought the symptoms were not significant and discharged the Pt who was brought back to KG the day after;

However, the symptoms occurred again. After a further dramatic phone conversation with our head, the parents accepted to bring the child to a further Hospital specialised in Pediatric Cardiology in Naples;

followed

The cardiologists accepted our suggestion and connected with our interventional cardiologists. Accordingly, they submitted the Pt to CC-A.

Outcome:subocclusion of the MT, LCA, RCA

Treatment: mismanaged (in our opinion)

Outcome: Obitus (occurred in a third Hospital in Rome; we tried to contact the Doctors even this time, but they denied collaboration)

followed

MT: mean trunk; LCA: left coronary artery; RCA: right coronary artery

Clinical case 1

Gender: F
Current age: 12

Genetic diagnosis: DHeFH

Age at diagnosis: 9

Baseline LDL-C (mg/dL): 553

E.O.:
Xanthomatosis: present
Arcus corneae: present
Xanthelasma: present

Clinical case 2

Gender: M
Current age: 11

Genetic diagnosis: HoFH

Age at diagnosis: 6

Baseline LDL-C (mg/dL): 609

E.O.:
Xanthomatosis: present
Arcus corneae: present
Xanthelasma: present

DHeFH: Double Heterozygous Familial Hypercholesterolemia; HoFH: Homozygous Familial Hypercholesterolemia

Clinical case 1

CV Diagnosis

Noninvasive
- ECG: normal
- Echocardiography: AVD
- Carotid echocolordoppler: normal

Invasive
Angiography: normal

Clinical case 2

CV Diagnosis

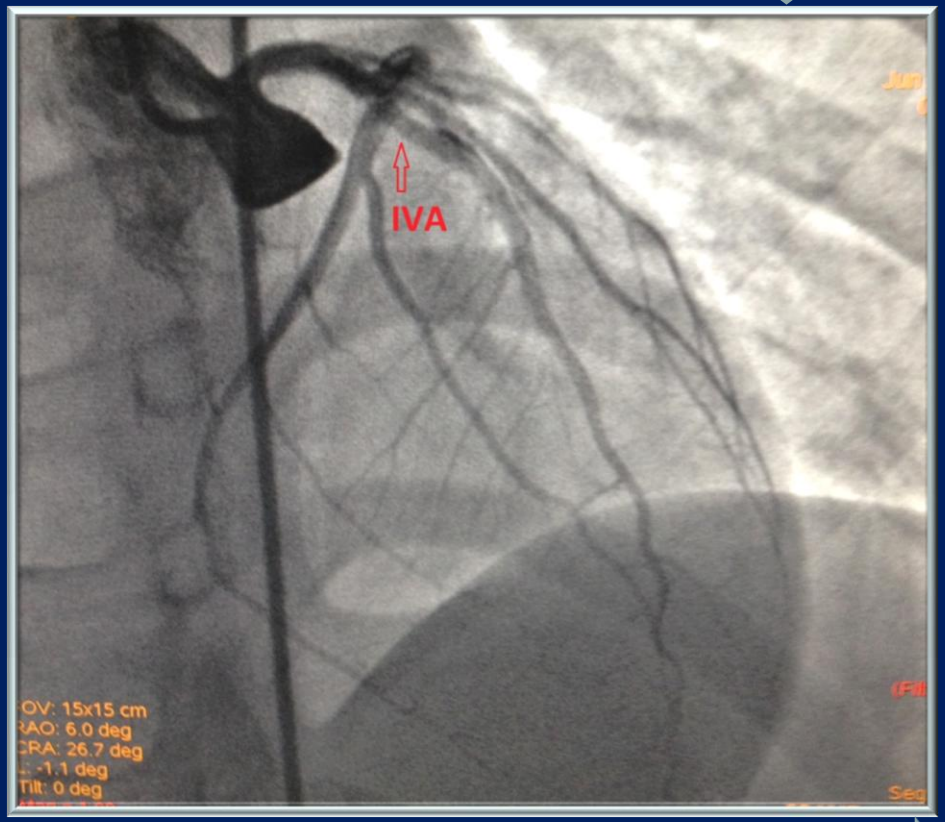
Noninvasive
- ECG: normal
- Echocardiography: normal
- Carotid echocolordoppler: normal

Invasive
Angiography: CAD

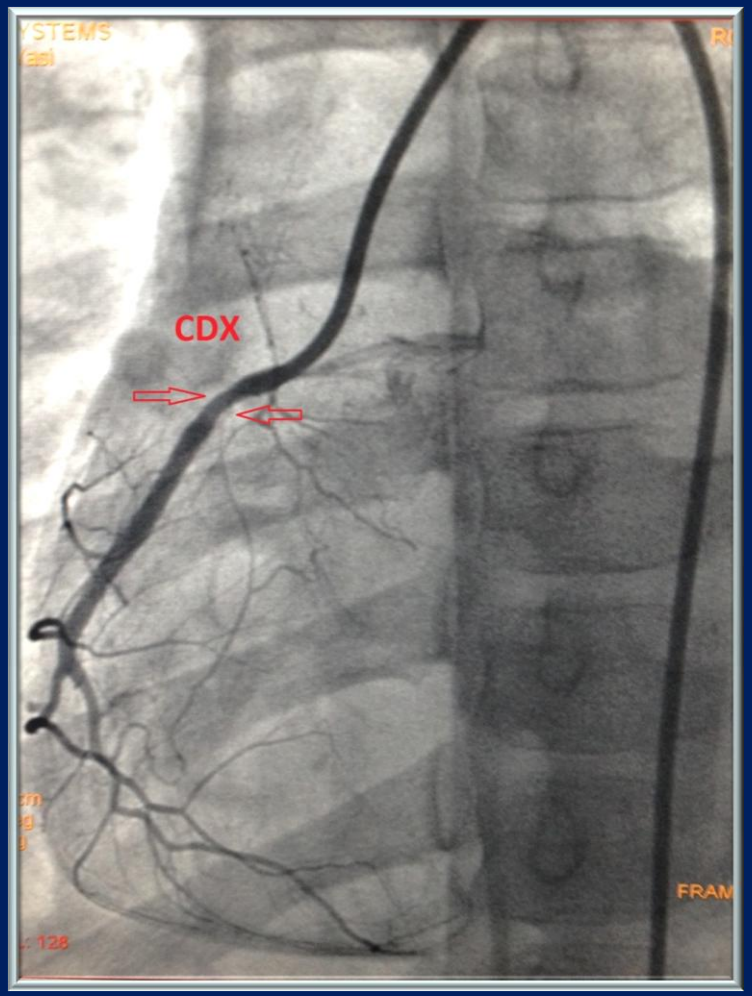
CV: Cardiovascular Diagnosis; CAD: Coronary artery disease; AVD: Aortic Valve Disease

RD ♂ 6 years; HoFH; Catheterisation of coronary arteries

Left coronary artery (LCA) (Anterior Intraventricular Branch) 20-30% stenosis



Right coronary artery (RCA), Proximal, 30% stenosis



Claudia Stefanutti, 2015

Clinical case 1

Treatment:
LA
Rosuvastatin 20 mg/d
Ezetimibe 10 mg/d
Diet Step II AHA
Physical Activity *

Age at initiation of LA: 9 years (02/07/14) after CVC placement (bilumen Tesio, right atrium and sup cava vein)

Method: Life18 by CVC

Frequency: every 10 days

***Strongly limited by CVC**

Clinical case 2

Treatment:
LA
Simvastatin 10 mg/d
Ezetimibe 10 mg/d
Diet Step II AHA
Physical Activity

Age at initiation of LA: 6 years (29/07/2013)

Method: Kaneka MA-03 - Liposorber - by peripheral venous access

Frequency: every 15 days

LA: Lipoprotein Apheresis; CVC: Central Venous Catheter

Clinical case 1

Outcomes:

Nurse at home every three days for CVC management;

01 Jul 14: mild aortic valve insufficiency (caused by catheter ??? As reported by the operator);

Jul 2014 - Dec 2014: hospitalization for repositioning of CVC with subsequent displacement and repositioning, several times;

Clinical case 2

Outcomes:

Clinical case 1

Outcomes:

Dec 2014: psychological counseling was begun;

19 Dec 14: hospitalization for sepsis and distal thrombus in CVC, resolved with antibiotic, fibrinolytic and anti-platelets therapies;

Jan 2015 - May 2015: 3 CVC replaced for granuloma;

Clinical case 2

Outcomes:

Clinical case 1

Outcomes:

May 2015: hospitalization for granuloma treated by silver nitrate pencils and local antibiotics. Insertion of a new CVC;

Dec 2015: hospitalization for staphylococcus aureus sepsis treated by ceftazidime and vancomycin plus urokinase;

25 May 2016: hospitalization for remove CVC and inflammatory tissue. Curettage of the insertion site. Replacing of new CVC;

Clinical case 2

Outcomes:

Clinical case 1

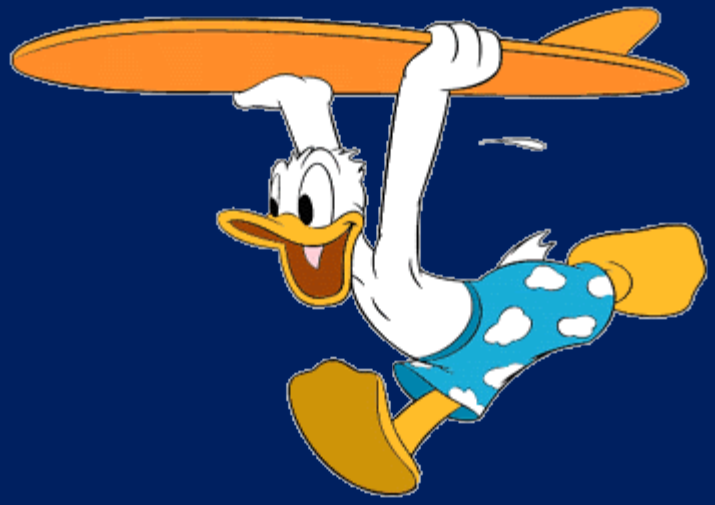
Outcomes:

15 Nov 2016: consulting at our centre and with parents approval we took the decision to remove the CVC;

05 Dec 2016: 1° LA procedure in our centre with Kaneka MA-03 - Liposorber - by peripheral venous access without any technical inconvenient (scheduled treatment frequency: 2QW).

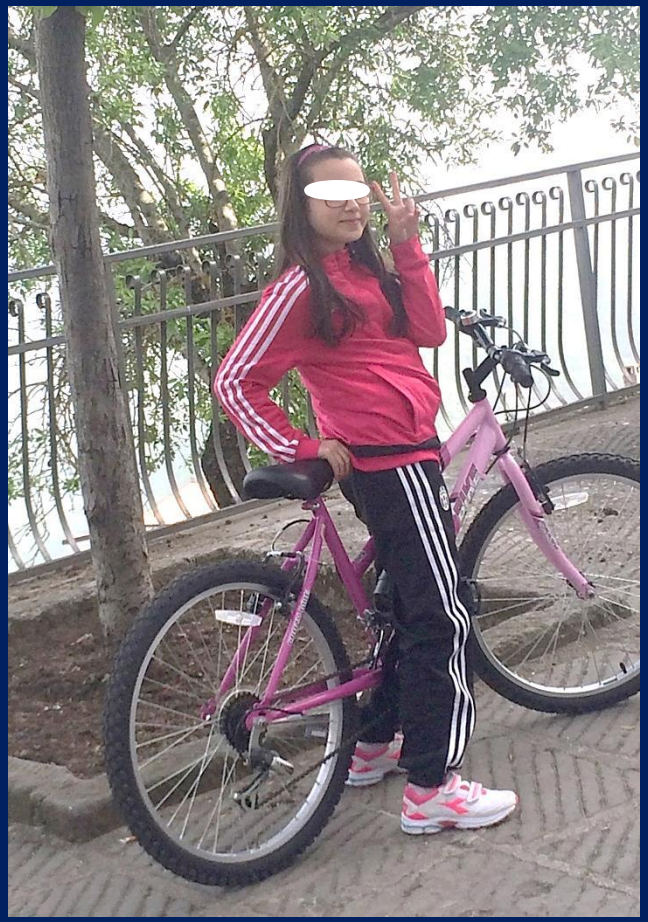
Clinical case 2

Outcomes:



Clinical case 1

Current status:



Clinical case 2

Current status:

Ph: ‘’Il Giornale di Latina’’, mercoledi 04 Nov 2015

Take Home Message

What do we learn from this two clinical cases?

- **In HoFH CV assessment must be undertaken prior to any attempt to treat**
- **Early treatment is mandatory**
- **CVC in HoFH children is not recommended and potentially harmful leading to severe complications**
- **Intrerdisciplinary consultation and collaboration is strongly recommended to avoid mismanagement of HoFH children**