PROPOSAL FOR A STANDARD EXAM FOR FH PATIENTS

1. History:

Family History:

- Focus on CVD incidence, ages of all first- and second-degree relatives at the time of the 1st CVD event / diagnosis, including sudden cardiac death
- Incidence of high cholesterol in family members; it is necessary to inquire about children and whether they have already been tested (and at what age)
- Incidence of FH-like findings found on physical examination relative to the family (incidence of eyelid xanthelasmas, tendinous xanthomas, etc. – including possible cosmetic or other surgeries for this reason)

Patient History:

- History of hypercholesterolemia incidence, possible previous treatment; it is necessary to determine previous undesirable side effects of treatment (myalgia, muscle weakness, elevated liver enzyme tests, memory disorders, etc.)
- Incidence of CVD in patients and age at the time of the 1st event / diagnosis
- Incidence of FH-like findings found on physical examination relative to the family (incidence of eyelid xanthelasmas, tendinous xanthomas, etc. – including possible cosmetic or other surgeries for this reason)
- Questions should exclude secondary causes of hypercholesterolemia (thyroid disease as well as other causes such as hormonal disorders, anorexia, hormonal contraceptive use, etc.).

Substance Abuse:

- Smoking – how many years, how many cigarettes per day, cessation attempts, whether the patient would like to quit

Subjective complaints:

Physiological complaints such as e.g., dyspnea NYHA III vs. never experienced dyspnea; experienced angina pectoris or CCS II vs. no limitations; lower extremity claudication; weight stability – or weight fluctuation (to what degree and for how long)

2. Objective evaluation:

- Blood pressure, height, weight, waist circumference, calculated BMI
- Standard physical examination focused on arcus lipoides corneae, xanthelasma palpebrarum, tendinous xanthomas, possible associated surgeries
- Murmurs over the aortic valve, over the carotid arteries, and over the large arteries of extremities
- Signs of peripheral arterial disease: unilaterally absent pulse, cold extremities, hairless, possible livid erythema, possible incidence of any distal necrotic changes
- ECG

3. Biochemical tests

- Initial tests: ALT, AST, TC, LDL, HDL, TG, apo B, apo A-I, lipoprotein(a), creatine kinase, glycemia, TSH
- Regular check-up tests: ALT, AST, TC, LDL, HDL, TG, creatine kinase, glycemia + others if possible
- Initial genetic testing: in the event of severe isolated hypercholesterolemia – LDL receptor mutations, and mutations in apolipoprotein B100
- In the event of elevated triglyceridemia: apolipoprotein E
- If genetic testing is not currently available, it is appropriate to draw a sample and freeze for future DNA analysis.

4. **Imaging exams** should be focused on verification of existing atherosclerotic processes.

- Carotid ultrasound (necessary if murmurs are heard above the carotid)
- ABI (ankle brachial index) appropriate for suspected peripheral arterial disease (incidence of claudication, possible clinical signs in the lower extremities)
- ECHO: in patients with severe isolated hypercholesterolemia to confirm or rule out degenerative affectations of the aortic valve, and to determine if myocardial ischemia is present
- Bicycle ergometer test and myocardial SPECT: for suspected myocardial ischemia
- Coronary calcium score
- Peripheral CT angiography
- Coronarography or angiography of peripheral arteries if there is a strong clinical suspicion of atherosclerotic disease