

Article

Diagnosis and screening strategies for detection of Familial Hypercholesterolaemia in children and adolescents in Italy: a survey from the LIPIGEN Paediatric Group

Supplementary Material

Survey

1. Indicate your affiliated LIPIGEN center:
2. In which region is your center located?
3. Is there a screening program for familial hypercholesterolemia in your region?
 - ☐ Yes, regional
 - ☐ Yes, local
 - ☐ No
4. The center where you operate is a clinic/specialized center for:
 - ☐ Both adults and pediatric patients
 - ☐ Exclusively pediatric patients
5. How many pediatric patients (under 18 years old) undergo a first visit at your center each year?
 - ☐ < 10
 - ☐ 10-20
 - ☐ 20-40
 - ☐ > 40
6. How many pediatric patients (under 18 years old) are currently in follow-up at your center?
 - ☐ < 50
 - ☐ 50-100
 - ☐ 100-200
 - ☐ > 200
7. What screening method for familial hypercholesterolemia is implemented for pediatric patients at your center?
(Multiple answers possible)
 - ☐ Universal screening
 - ☐ Selective screening (for a specific high-risk population)
 - ☐ Cascade screening (from an adult index case to family members, including children/adolescents)
 - ☐ Reverse screening (from a pediatric index case to other family members)
 - ☐ Child-parent screening (from a child screened at a predetermined age to the parent)
 - ☐ Other (please, specify...)
8. In your opinion, what is the most appropriate screening method for familial hypercholesterolemia?
 - ☐ Universal screening
 - ☐ Selective screening
 - ☐ Cascade screening

- Reverse screening
 - Child-parent screening
 - A combination of different methods
 - Other (please, specify...)
9. In the diagnostic process for pediatric patients with hypercholesterolemia, do you routinely apply cascade screening?
- Always
 - 50%
 - < 50%
 - Never
10. In the diagnostic process for pediatric patients with hypercholesterolemia, do you routinely apply reverse screening?
- Always
 - 50%
 - < 50%
 - Never
11. At your center, which biochemical/genetic parameters are requested for the assessment of familial hypercholesterolemia?
- (Multiple answers possible)*
- Lipid profile: Total Cholesterol (TC), HDL-C, LDL-C, Triglycerides (TG)
 - Lipoprotein profile: ApoA1, ApoB
 - Lp(a)
 - Genetic analysis
12. At your center, is Lp(a) evaluation requested for the assessment of pediatric patients with familial hypercholesterolemia?
- Always
 - 50%
 - < 50%
 - Never
13. At your center, in cases of clinical suspicion, is genetic analysis requested for the diagnosis of familial hypercholesterolemia in pediatric patients?
- Always
 - 50%
 - < 50%
 - Never
14. What are the main criteria you use to request genetic analysis for pediatric patients?
- (Multiple answers possible)*
- LDL-C levels
 - Family history of cardiovascular disease (CVD)
 - Family member with hypercholesterolemia
 - Poor response to dietary intervention

15. Which physicians most commonly refer pediatric patients to your center?

(Multiple answers possible)

- ☐ Primary care pediatricians
- ☐ General practitioners
- ☐ Cardiologists
- ☐ Other:

16. What percentage of pediatric patients (under 18 years old) undergoing a first visit at your center are referred by a primary care pediatrician/general practitioner after detecting hypercholesterolemia?

- ☐ < 10%
- ☐ 10-25%
- ☐ 25-50%
- ☐ 50-75%
- ☐ > 75%

17. What percentage of pediatric patients (under 18 years old) undergoing a first visit at your center are referred by a general practitioner treating a parent or first-degree relative with hypercholesterolemia (cascade screening)?

- ☐ < 10%
- ☐ 10-25%
- ☐ 25-50%
- ☐ 50-75%
- ☐ > 75%

18. What percentage of pediatric patients (under 18 years old) undergoing a first visit at your center are referred on the initiative of a family member with hypercholesterolemia?

- ☐ < 10%
- ☐ 10-25%
- ☐ 25-50%
- ☐ 50-75%
- ☐ > 75%

19. You may leave a comment:

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