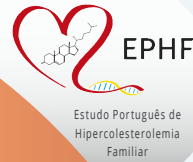




Familial Hypercholesterolemia

Portuguese population with a clinical diagnosis of FH

1999-2024 (Last updated on 31-12-2024)



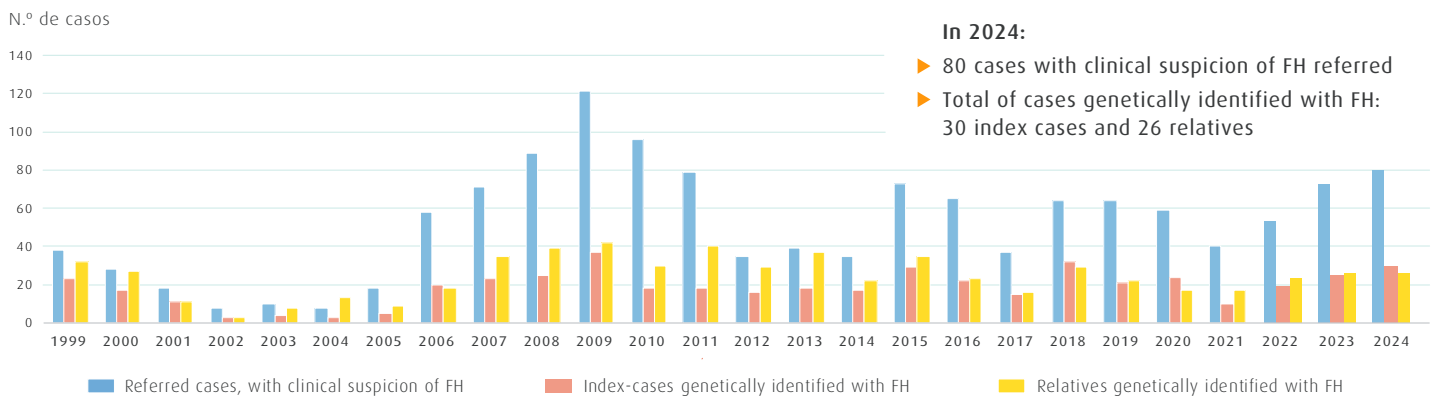
Since 1999, INSA has been coordinating the Portuguese Familial Hypercholesterolemia Study. This study allows the identification of the genetic cause of hypercholesterolemia (high cholesterol levels since birth) in a family, in order to reduce their cardiovascular risk, with early diagnosis and adequate treatment.

- ▶ Familial Hypercholesterolemia (FH) is a common genetic disorder, and people with FH have a high cardiovascular risk.
- ▶ In Portugal, there are approximately 33.000 people with heterozygous FH and 25 with homozygous FH.
- ▶ Studies indicate that FH is underdiagnosed and identified late, after the age of 40 and only 2% before the age of 18.
- ▶ In Portugal, only 3% of people with FH are identified.

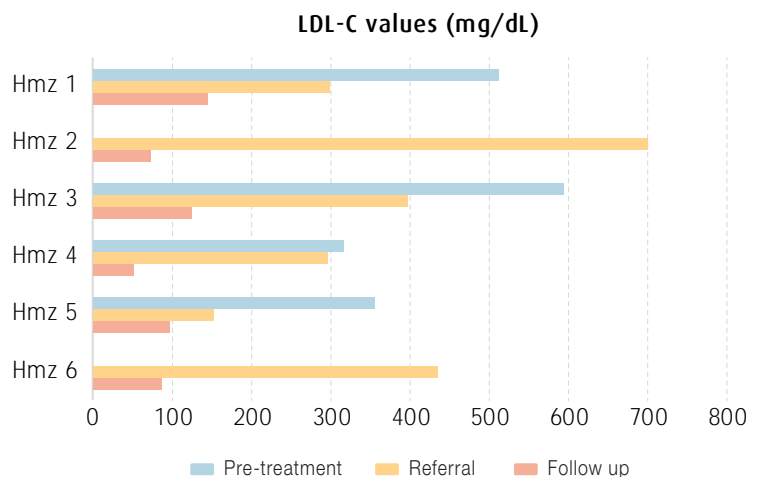
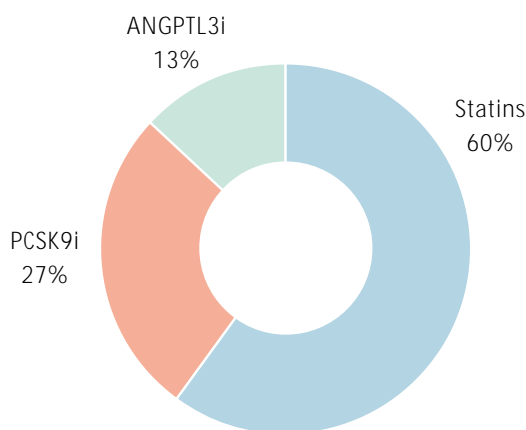
- ▶ National pediatric screening programs are essential to identify individuals with FH early in life.
- ▶ With the availability of novel therapies for children, FH has become a well treatable disease.
- ▶ Early detection and treatment of children with FH not only is health-saving, but also cost-effective.
- ▶ Pediatric screening will benefit the entire family, as it will allow the identification of at least the affected parent, and also all affected relatives.

Between 1999 and 2024, 3775 cases were referred, of which 1386 index cases with clinical suspicion of FH, and 2389 relatives.

Total of cases genetically identified with FH: 1150 (1129 heterozygous FH + 15 homozygous FH).

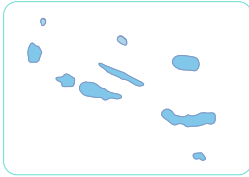


15 homozygous identified between 1999-2024
40% taking novel therapies (follow up data)



Percentage of cases genetically identified with FH, by region
(considering a prevalence of 1:300)

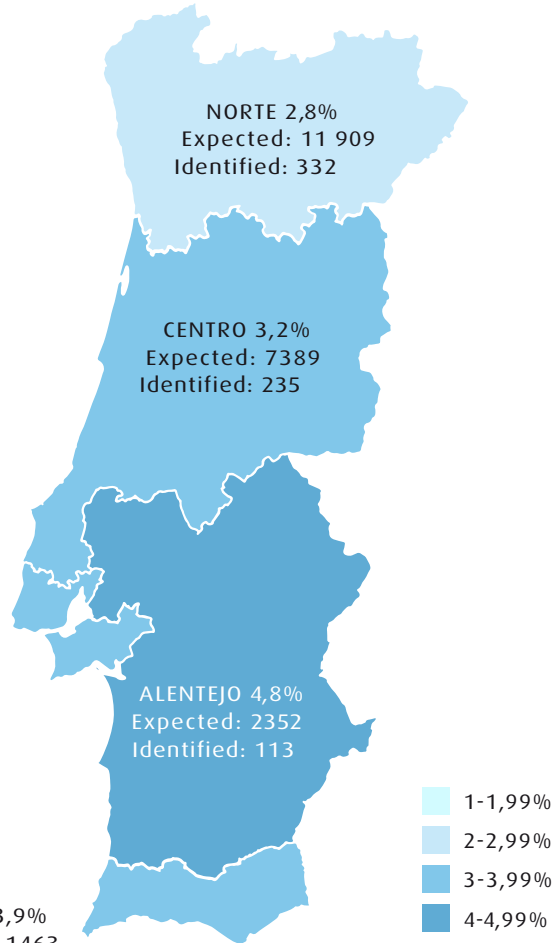
R.A. AÇORES 3%
Expected: 809
Identified: 24



R.A. MADEIRA 1,3%
Expected: 846
Identified: 11

ÁREA METROPOLITANA DE LISBOA 3,2%
Expected: 9488
Identified: 311

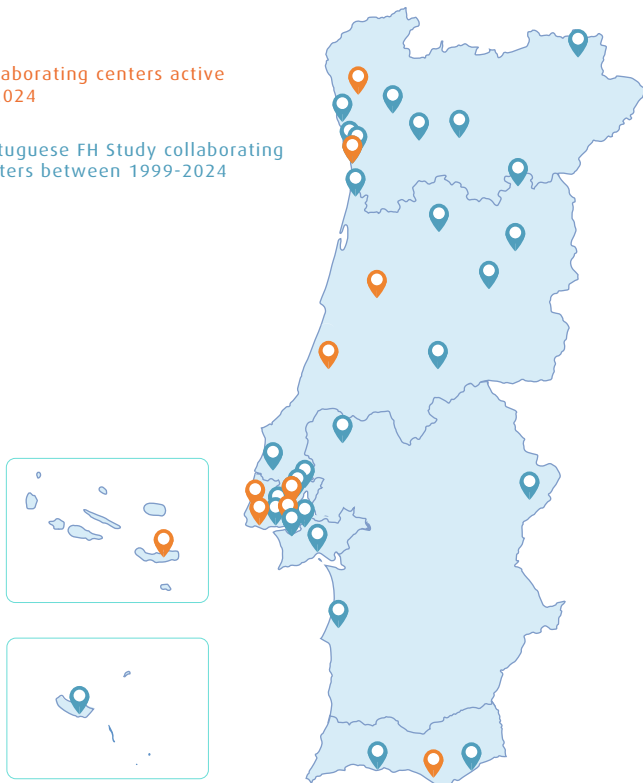
ALGARVE 3,9%
Expected: 1463
Identified: 57



Collaborating institutions of the Portuguese FH Study

Collaborating centers active in 2024

Portuguese FH Study collaborating centers between 1999-2024



BRAGANÇA: C S Bragança, Medicina Geral e Familiar; Unidade Saúde Local do Nordeste, Pediatria.
VILA DO CONDE: C H Póvoa Varzim - Vila do Conde, Medicina Interna. **BARCELOS:** H Sta Maria Maior, Medicina Interna e Pediatria. **BRAGA:** H de Braga, Pediatria. **GUIMARÃES:** H Da Senhora da Oliveira - Guimarães, Cardiologia e Pediatria. **SENHORA DA HORA:** H Pedro Hispano, Cardiologia e Medicina Interna. **PORTO:** C H e Univ do Porto - H Sto António, Endocrinologia e Pediatria; C H e Univ do Porto - Maternidade Júlio Dinis, Genética Médica; Centro Materno Infantil do Norte, Pediatria; H S João, Genética Médica, Cardiologia e Pediatria. **VILA NOVA DE GAIA:** H da Luz Arrábida, Cardiologia. **STA MARIA DA FEIRA:** C H de Entre o Douro e Vouga - H S Sebastião, Pediatria. **VILA REAL:** C H de Trás-os-Montes e Alto Douro, Pediatria. **AMARANTE:** UCSP Figueiró, Medicina Geral e Familiar; **UISEU:** C H Tondela - Viseu, Pediatria. **GUARDA:** UCSP Guarda - Extensão S Miguel, Medicina Geral e Familiar. **MEDA:** C S de Meda, Medicina Geral e Familiar. **COVILHÃ:** C H Cova da Beira - Covilhã, Medicina Interna e Pediatria. **COIMBRA:** C H e Univ de Coimbra, Medicina Interna; H Pediátrico Carmona da Mota, Cardiologia Pediátrica, Genética Médica e Pediatria. **PROENÇA-A-NOVA:** C S Proença-a-Nova, Medicina Geral e Familiar. **LEIRIA:** C H Leiria Pombal - H Sto André, Medicina Interna, Endocrinologia e Pediatria. **SANTARÉM:** H Santarém, Pediatria. **TORRES VEDRAS:** C H Oeste - Hospital Torres Vedras, Medicina Interna. **VILA FRANCA DE XIRA:** H Vila Franca de Xira, Pediatria. **LOURES:** C S Unhos, Medicina Geral e Familiar; H Beatriz Ângelo, Pediatria e Medicina Interna. **AMADORA:** H Prof. Doutor Fernando Fonseca, Medicina Interna e Pediatria. **LISBOA:** Clínica CUF Belém, Medicina Geral e Familiar; H Cruz Vermelha, Pediatria; H CUF Descobertas, Medicina Geral e Familiar; H Curry Cabral, Endocrinologia; H D Estefânia, Pediatria; H da Luz, Cardiologia e Medicina Interna; H Egas Moniz, Endocrinologia e Genética Médica; H Lusíadas Lisboa, Medicina Interna e Pediatria; H Militar, Cardiologia; H Pulido Valente, Cardiologia; H Sta Marta, Cardiologia, Cardiologia Pediátrica e Medicina Interna; H Sta Maria, Cardiologia, Genética Médica, Medicina Interna e Pediatria; Instituto Nacional de Cardiologia Preventiva Prof Fernando de Pádua, Genética Médica; USF das Conchas, Medicina Geral e Familiar. **ALGÉS:** USF Linha de Algés, Medicina Geral e Familiar. **CARNAXIDE:** H Sta Cruz, Genética Médica, Cardiologia, Cardiologia Pediátrica e Nefrologia. **CASCAIS:** H Cascais, Cardiologia, Medicina Interna e Pediatria. **SINTRA:** H CUF Sintra. **ALVERCA DO RIBATEJO:** Clínica Missão Saúde, Medicina Geral e Familiar. **ALMADA:** H Garcia da Orta, Cardiologia e Medicina Interna. **SEIXAL:** C S do Seixal, Medicina Geral e Familiar. **BARREIRO:** C H Barreiro Montijo, Pediatria. **SETÚBAL:** C H Setúbal, Cardiologia; H de Santiago, Cardiologia e Pediatria. **ELVAS:** H Sta Luzia De Elvas, Cardiologia. **SANTIAGO DO CACÉM:** H Litoral Alentejano, Medicina Interna e Pediatria. **LAGOA:** C S de Lagoa, Medicina Geral e Familiar. **FARO:** C H Algarve - Faro, Cardiologia e Pediatria. **TAVIRA:** CS Tavira, Medicina Geral e Familiar; **PONTA DELGADA:** H Divino Espírito Santo de Ponta Delgada, Endocrinologia, Endocrinologia Pediátrica, Medicina Interna e Cardiologia. **FUNCHAL:** H Marmeleiros, Medicina interna; H Dr Nélio Mendonça - Funchal, Pediatria