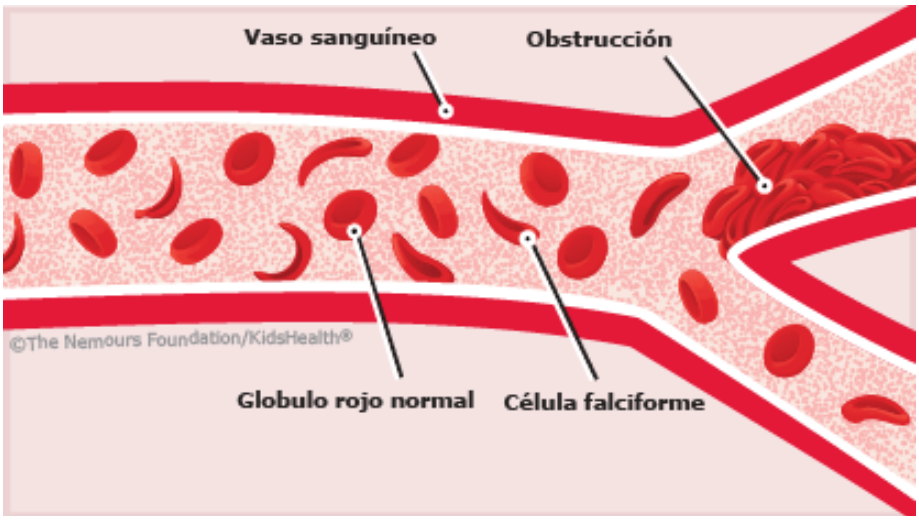
Sickle cell anemia as a model for the understanding and diagnosis of genetic disease



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IDEA INICIAL

- Hypothesis 1. We can predict that a mutation in the hemoglobin protein will produce sickle cell disease.

- Hypothesis 2. We can predict what mutations will cause inherited diseases.

- Hypothesis 3. Bioinformatics tools can be used as a support for disease diagnosis.

PROCÉS D'ELABORACIÓ

The bulk of the data was obtained from web searches and literature consultations. This information was complemented with two visits to centers of biomedical research (VHIR and IBMB-CSIC) and an interview to a specialized researcher. I obtained a practical feeling of the research by doing a molecular biology experiment at the IBMB-CSIC.

CONCLUSIONS

First, I have to say that all my hipothesis ended up being true.

At the academic level, I have learned the main principles underlying information retrieval, structuring and presentation. In this part I am particularly grateful to the support and guidance of my supervisor Silvia Flores, to the effort of all the teachers I have had and to Maristes Sants-Les Corts for giving me such a good academic environment.

At the scientific level, I have improved my skills about what is a scientific fact and about how it can be transformed into actual, useful knowledge. Particularly, I have learned a lot about the molecular basis of disease and how bioinformatics tools can be used to sort diagnosis problems.

In summary, developing this project has been for me a powerful learning experience that will undoubtedly leave a trace in my view of studies and professional work. I am grateful for this.