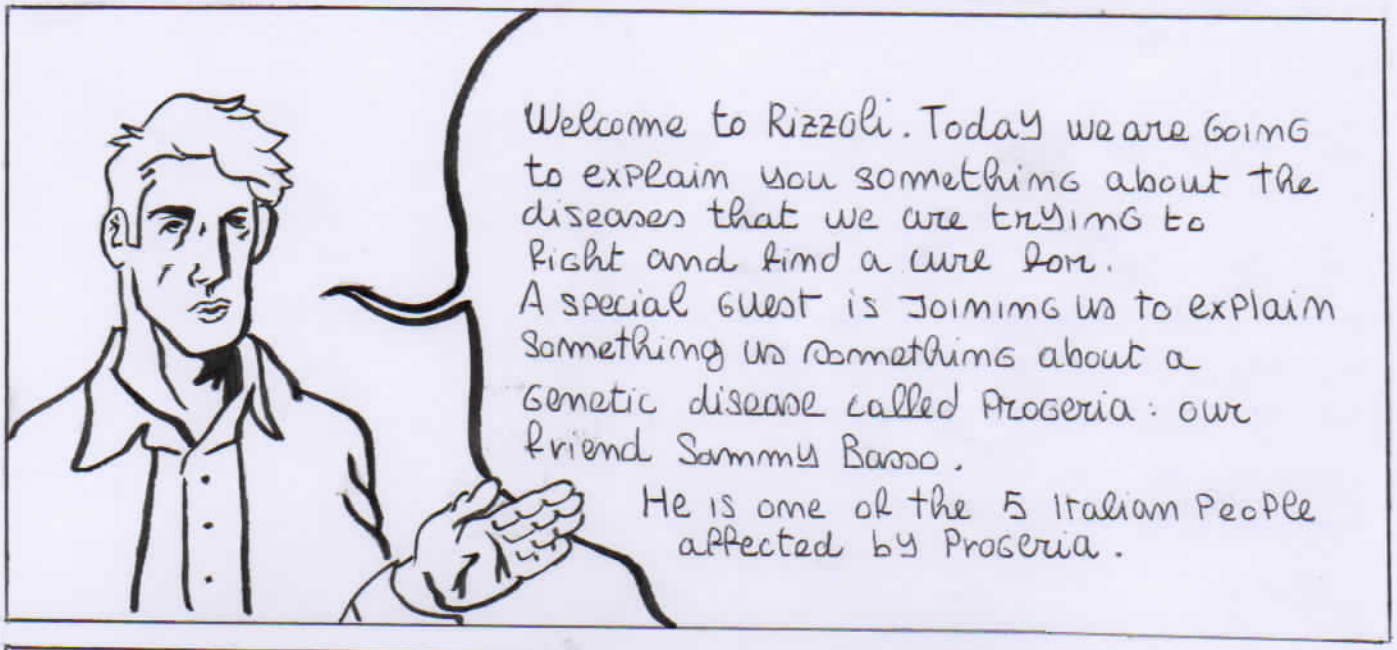
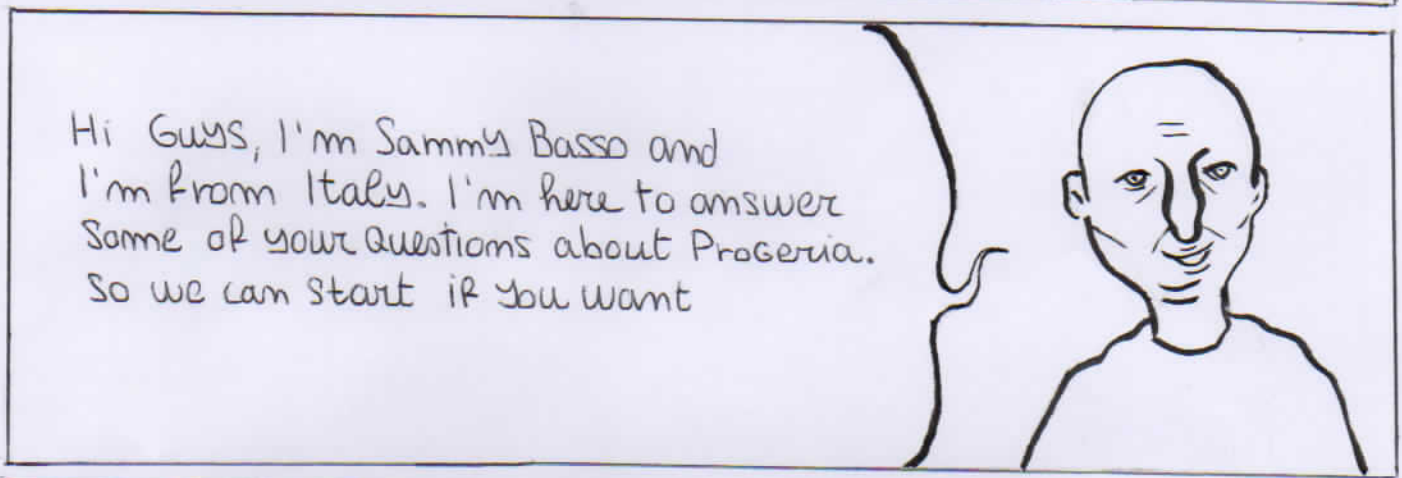
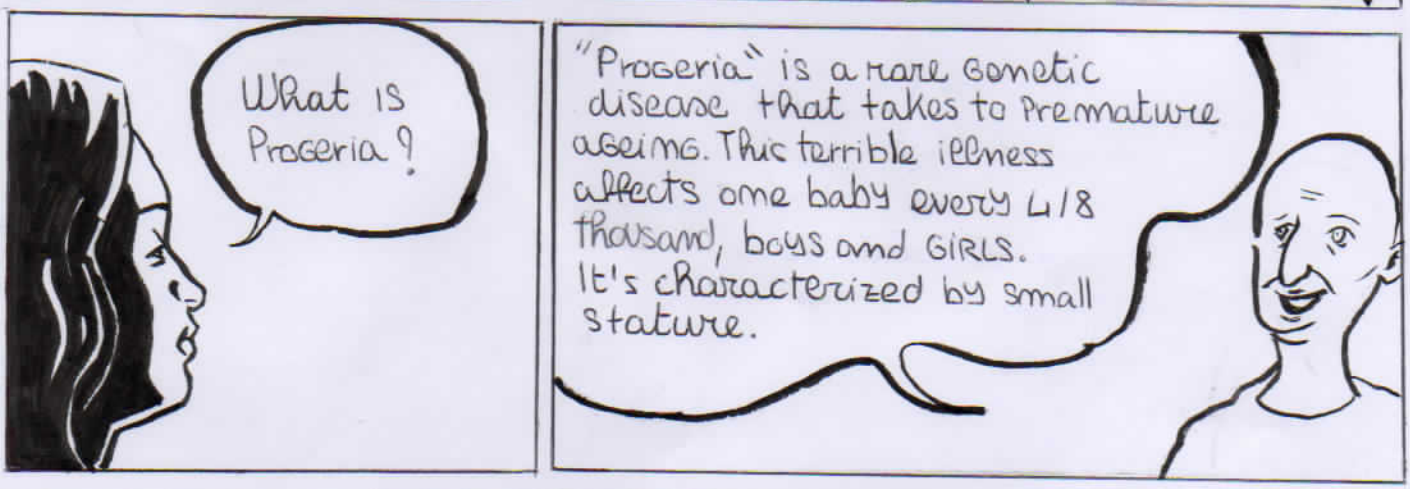
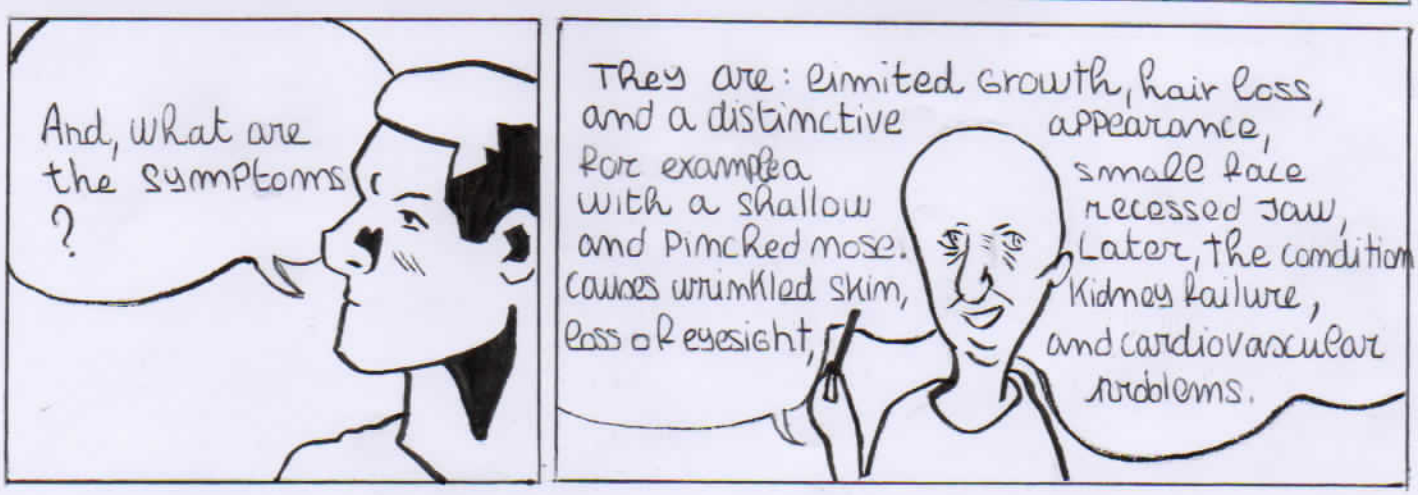
SAMMY'S WORLD


Welcome to Rizzoli. Today we are Going to explain you something about the diseases that we are trying to fight and find a cure for.
A special Guest is Joining us to explain Something us Something about a Genetic disease called Progeria: our friend Sammy Basso.
He is one of the 5 Italian People affected by Progeria.


Hi Guys, I'm Sammy Basso and I'm from Italy. I'm here to answer Some of your Questions about Progeria. So we can Start if you want


What is Progeria?
"Progeria" is a rare Genetic disease that takes to Premature ageing. This terrible illness affects one baby every 4/8 thousand, boys and GIRLS. It's characterized by small stature.


And, what are the symptoms?
They are: limited Growth, hair loss, and a distinctive appearance, for example with a shallow and pinched nose. small face recessed jaw, Causes wrinkled skin, Later, the condition loss of eyesight, kidney failure, and cardiovascular problems.

How does it work?

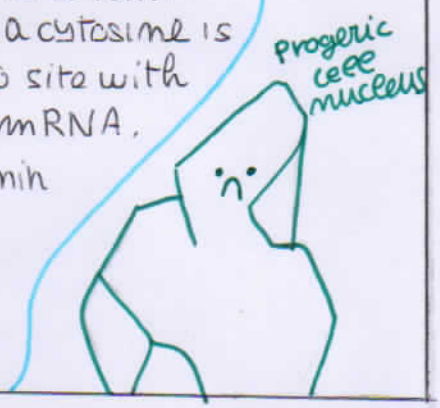
Progeria, or Hutchinson-Gilford syndrome, is a rare disease that causes premature aging, even though it doesn't affect the human mind, which is the only sign of the actual age of the person affected. It causes in the child the typical illnesses caused by age and it takes the person to an early death.
Progeria is a genetic disease that happens because of a de novo mutation and it's rarely hereditary.
It's caused by a sporadic mutation during the first phases of the embryonic developement

In human cells, the nucleus is delimited by the nuclear envelope, which consists of an outer nuclear membrane (structurally and functionally similar to the membrane of endoplasmic reticulum), an internal nuclear membrane, and the nuclear membrane.

In mammals there are at least 7 types of laminal that are coded by 3 genes (A, B, C)
The gene involved in the disease is the LMNA, wich encodes the prelamin A

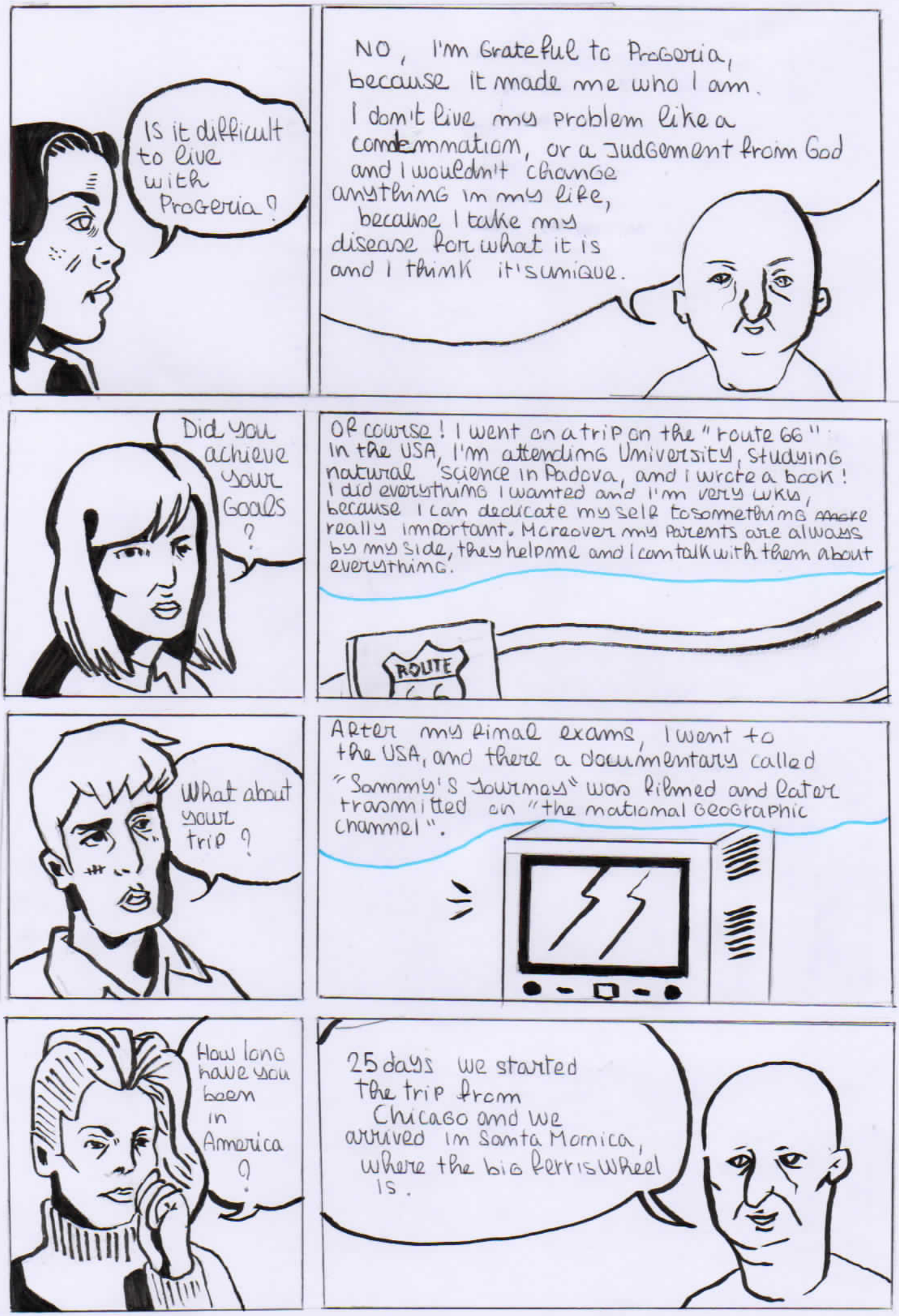
In 2003, it was discovered that the cause of Progeria is a point mutation in position 1824 of the LMNA gene, where a cytosine is replaced by a timin. This mutation creates a splicing site with exon 11, resulting in a brief and abnormal mature mRNA.
During translation, an abnormal variant of the Prelamin protein is producted.

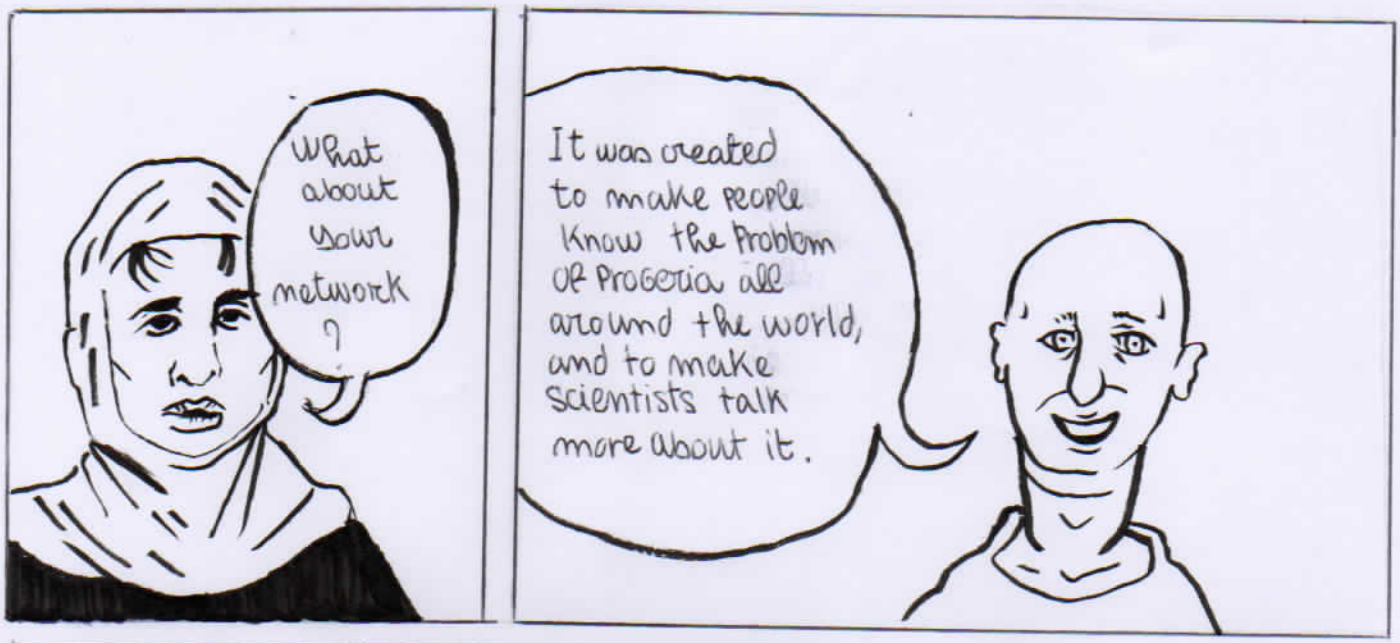
Unlike "accelerated aging diseases" (such as Werner syndrome or pigmentous xeroderma), Progeria is not caused by defective DNA repair.



Is there a cure for this disease?

No treatment has yet proven effective.
Most treatment options have focused on reducing complications with surgeries, but as there is no known cure, few people with Progeria go over 13 years of age.


Is it difficult to live with Progeria?
NO, I'm Grateful to Progeria, because it made me who I am. I don't live my problem like a condemnation, or a Judgement from God and I wouldn't change anything in my life, because I take my disease for what it is and I think it's unique.
Did you achieve your Goals?
OF course! I went on a trip on the "route 66" in the USA, I'm attending University, studying natural science in Padova, and I wrote a book! I did everything I wanted and I'm very lucky, because I can dedicate myself to something ~~make~~ really important. Moreover my parents are always by my side, they help me and I can talk with them about everything.
ROUTE
What about your trip?
After my final exams, I went to the USA, and there a documentary called "Sammy's Journey" was filmed and later trasmitted on "the national Geographic channel".
How long have you been in America?
25 days we started the trip from Chicago and we arrived in Santa Monica, where the big ferris Wheel is.



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