CENTER FOR NEUROSCIENCE AND CELL BIOLOGY

A RARE JOURNEY

A PROJECT FROM THE CENTER FOR NEUROSCIENCE AND CELL BIOLOGY OF THE UNIVERSITY OF COIMBRA, PORTUGAL

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SOME DISEASES ARE COMMON, SOME NOT AT ALL. THESE ARE CALLED **RARE**, OR **ORPHAN DISEASES**, AND HAVE A PREVALENCE OF LESS THAN 5 IN 10 000 PEOPLE.

IT IS ESTIMATED THAT THERE ARE 5 TO 8 000 DIFFERENT RARE DISEASES, AFFECTING UP TO 6% OF THE POPULATION, WHICH MEANS THAT THERE ARE UP TO 600 000 PEOPLE WITH THESE DISEASES IN PORTUGAL.

MACHADO-JOSEPH DISEASE IS A RARE DISEASE THAT WAS CHARACTERIZED IN THE 1970S-80S BY TWO PORTUGUESE RESEARCHERS, WHO STUDIED AZOREAN FAMILIES, SOME WITH DESCENT IN THE USA.

CORINO DE ANDRADE

PAULA COUTINHO

TWO OF THESE FAMILIES (MACHADO AND JOSEPH) NAMED THE DISEASE.

IN FACT, IN THE AZORES ARCHIPELAGO (AND ESPECIALLY IN THE FLORES ISLAND) THIS DISEASE IS NOT SO RARE.

1 IN 100000

AZORES ARCHIPELAGO

CORVO

FLORES

GRACIOSA

SÃO JORGE

TERCEIRA

FAIAL

PICO

SÃO MIGUEL

SANTA MARIA

1 IN 158 PEOPLE

THE **FLORES ISLAND** HAS THE HIGHEST PREVALENCE OF THE DISEASE IN THE WORLD.

MACHADO-JOSEPH DISEASE IS A **NEURODEGENERATIVE**, **HEREDITARY**, **PROGRESSIVE** AND **DOMINANT** DISEASE.

A CHILD WITH ONE DISEASED PARENT IS 50% LIKELY TO HAVE IT, AND SYMPTOMS MANIFEST MOSTLY WHEN PATIENTS ARE BETWEEN 35 AND 50 YEARS OLD.

THE PROBLEM OF THIS PATHOLOGY LIES IN A CHANGE IN A SPECIFIC GENE, WHICH AFFECTS THE FUNCTION OF CERTAIN BRAIN AREAS: THE **CEREBELLUM**, THE **BRAINSTEM**, THE **BASAL GANGLIA** AND THE **UPPER PART OF THE SPINAL CORD**.

CEREBELLUM

BRAINSTEM

SPINAL CORD

THESE BRAIN STRUCTURES ARE RESPONSIBLE FOR **BALANCE** AND **MOVEMENT**.

THEREFORE, MACHADO-JOSEPH PATIENTS HAVE PROGRESSIVELY IMPAIRED MOVEMENT COORDINATION, AND LOSS OF BALANCE (THE SO-CALLED "**ATAXIA**").

WHICH CAN CULMINATE IN GAIT AND BALANCE ISSUES AND PROBLEMS IN SPEAKING, SWALLOWING, EYE MOVEMENT AND SLEEP.

1, 2, 3...

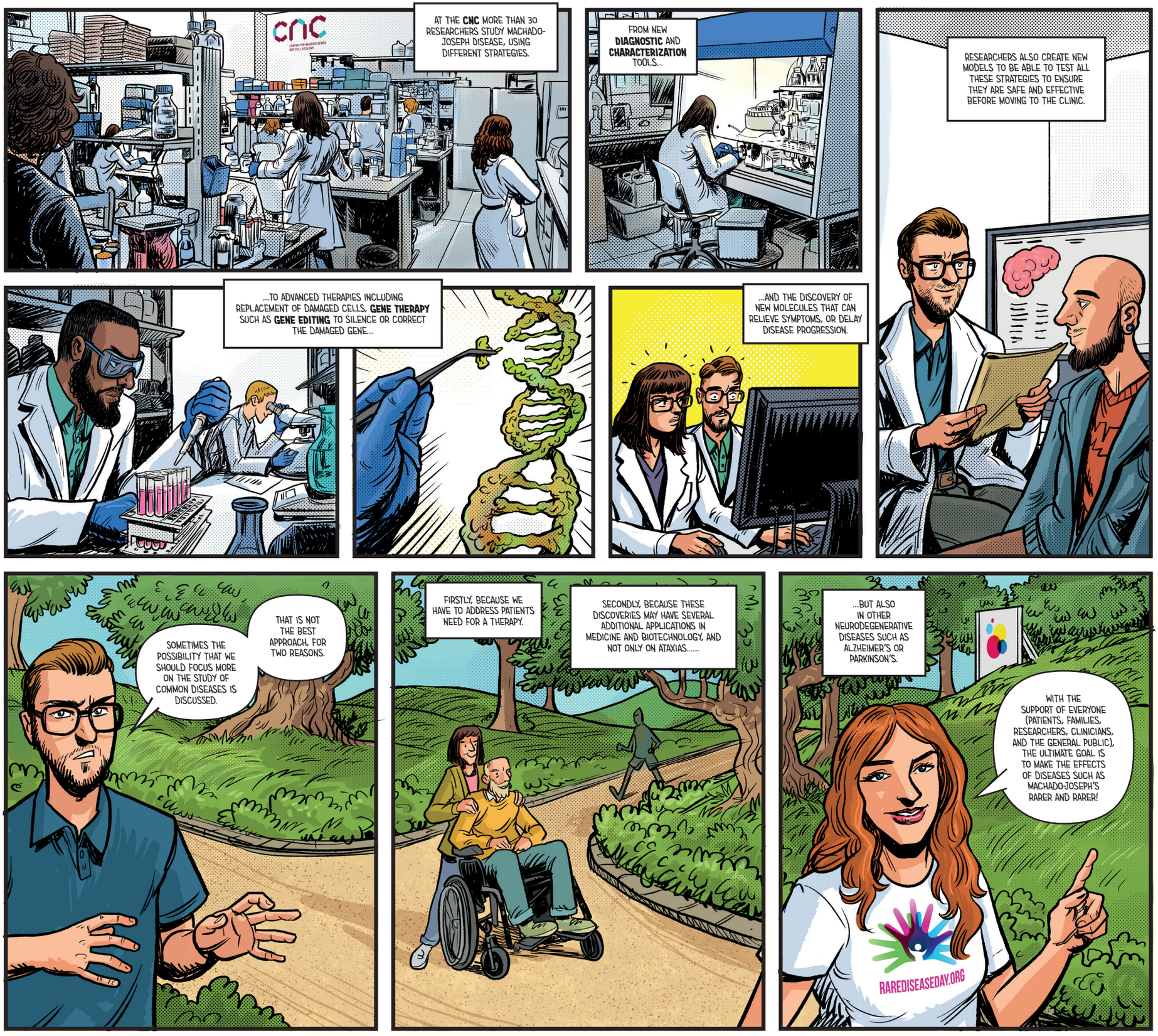
THE OTHER NAME BY WHICH THIS DISEASE IS ALSO KNOWN, **TYPE 3 SPINOCEREBELLAR ATAXIA**, RESULTS FROM BOTH THE SYMPTOMS AND BRAIN AREAS INVOLVED.

MACHADO-JOSEPH IS ONE OF THE MOST COMMON GENETIC-BASED ATAXIAS, BUT THERE ARE MANY SIMILAR DISEASES THAT ARE ALSO CLASSIFIED AS "ATAXIAS".

UNFORTUNATELY, THIS DISEASE CURRENTLY HAS NO CURE. PHYSIOTHERAPY...

...AND SOME MEDICATION HELPS REDUCE SYMPTOMS.

BUT THERE IS HOPE, BOTH IN RESEARCH AND IN ONGOING CLINICAL TRIALS.



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