Method and kit for the rapid sequencing of the human ATM gene and method for the diagnosis of Ataxia Telangiectasia.

KEYWORDS

- ATM
- PLATE SEQUENCING
- POINT MUTATIONS
- RAPID PROTOCOL
- COST SAVING

AREA

- BIOMEDICAL

Priority Number
n. 102015000051619 _ 15.09.2015.

Patent Type
Patent for invention.

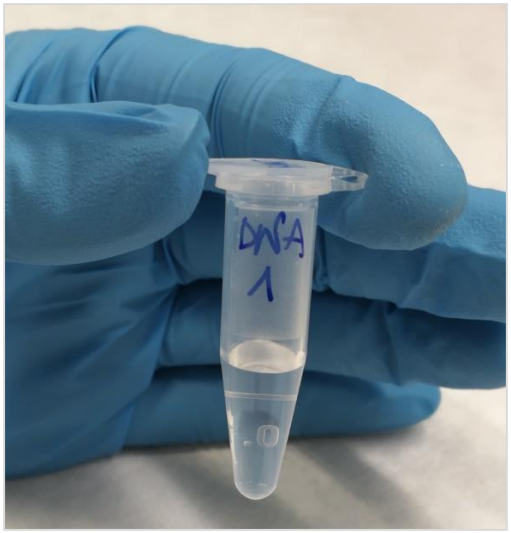
Ownership
Sapienza University of Rome 100%.

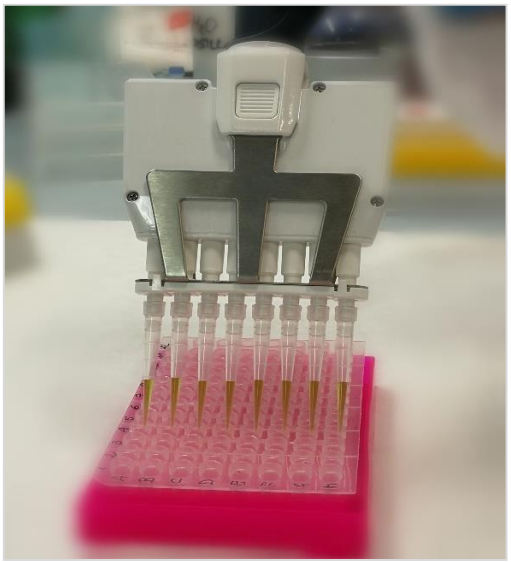
Inventors
Camilla Savio, Luciana Chessa.

Industrial & Commercial Reference
Biomedical companies, molecolar biology kit marketing.

Time to Market
The entire procol was tested on DNA sample previously characterized to verify the results. Perfect matching with classical protocol.

Availability
Licensing and Collaboration.





Abstract
The ATM gene is responsible for autosomal recessive pathology Atassia Telangiectasia (AT) but healthy carriers of a mutation are more susceptible to cancers of the normal population.

The gene is very extensive and point mutations (about 80% of all mutations) may be scattered throughout the gene exons.

The object of the invention is a method, a rapid process for simultaneous sequencing of all gene exons in order to find any mutations for confirmation of AT clinical diagnosis and for carriers.

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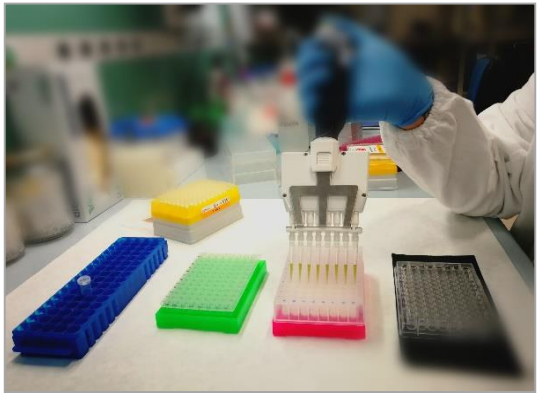
Method and kit for the rapid sequencing of the human ATM gene and method for the diagnosis of Ataxia Telangiectasia.

Technical Description

The kit consists of 65 pairs of primers to amplify the entire gene and distributed in a multi-well plate to undergo a single simultaneous amplification (PCR) process.

The process further provides that the subsequent steps of purification and sequence reaction are carried out in the plate using a multichannel pipette.

The distribution of primers by sequence also follows a specific scheme so to not alter the order of the amplified exons in the plate.
It is therefore possible to sequencing the whole ATM gene with few and simultaneous steps so that time and costs are greatly reduced.

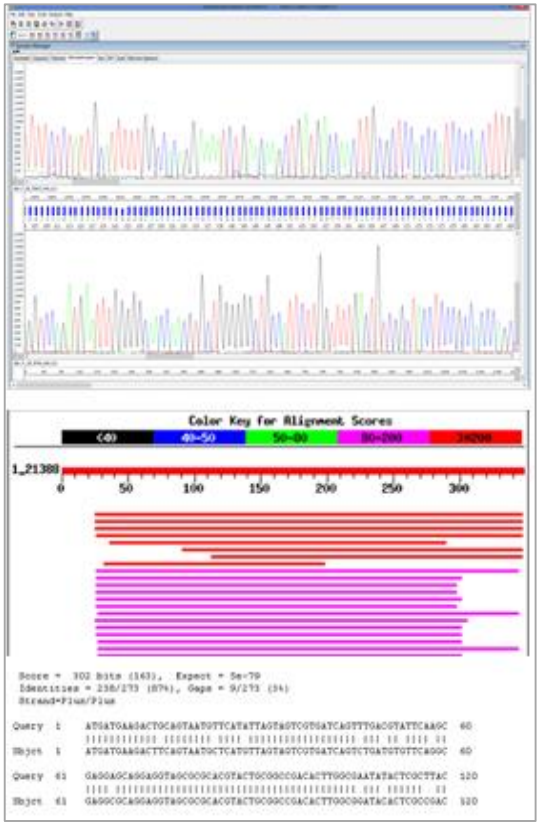


Technologies & Advantages

The process described above makes it possible to perform sequencing very quickly, because it allows simultaneous preparation of all exons of the gene to be analyzed. It reduces the possibility of error, since the primers are already distributed in the plate and a plate is used to analyze a single sample. It is a much cheaper system than the single preparation of each exon to be sequenced. It does not require additional techniques to be validated in comparison to NGS for example.

Applications

Application in biomedical field and specifically in molecular diagnostics and research laboratories.



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