CASE STUDY

Analyzing patient samples with mass spectrometry genotyping

This case study was presented by Dr. Elisa Chiadini from Bioscience Laboratory of IRST-IRCCS in Meldola, Italy.

THE SITUATION

- Patient samples were analyzed for lung and colon cancer using direct sequencing or pyrosequencing.
- Workflow duration was very long as parallel processing was impossible and not all clinically relevant mutations could be detected (in the case of faster pyrosequencing).
- Frequent repetition was required due to considerable background noise.
- Sensitivity was low, which required subsequent nested PCR, introducing a series of biases into the workflow.
- Patient reports could not be delivered in a timely manner and treatment of patients with targeted cancer drugs was delayed.

THE CHALLENGE

- Establish a workflow able to detect all clinically relevant mutations in a timely manner and with sufficient sensitivity and specificity.
- Avoid repetitions and/or re-analysis with further methods.

THE SOLUTION

- Using MassARRAY® Dx technology from Agena Bioscience, samples for several alterations could be analyzed in a single workflow. Results were easy to interpret and assay repetitions were rarely necessary.
- Minimal input DNA was required, as opposed to sequencing and pyrosequencing, ensuring samples of poor quality or small size could be analyzed.
- High sensitivity using the Myriapod® Lung Status enabled detection of mutations with frequencies as low as 5%. Lung and colon cancer status kits detected a large number of genes for every analyzed sample.
- Oncologists received high quality information on a patient's cancer at a faster rate, and in time for a swift initiation of targeted therapy.

FURTHER BACKGROUND INFORMATION

- The information on markers provided by the MassARRAY Dx technology may also be useful to choose future targeted therapies.

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Learn more about MassARRAY Dx.
http://agenabio.com/products/massarray-dx/

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