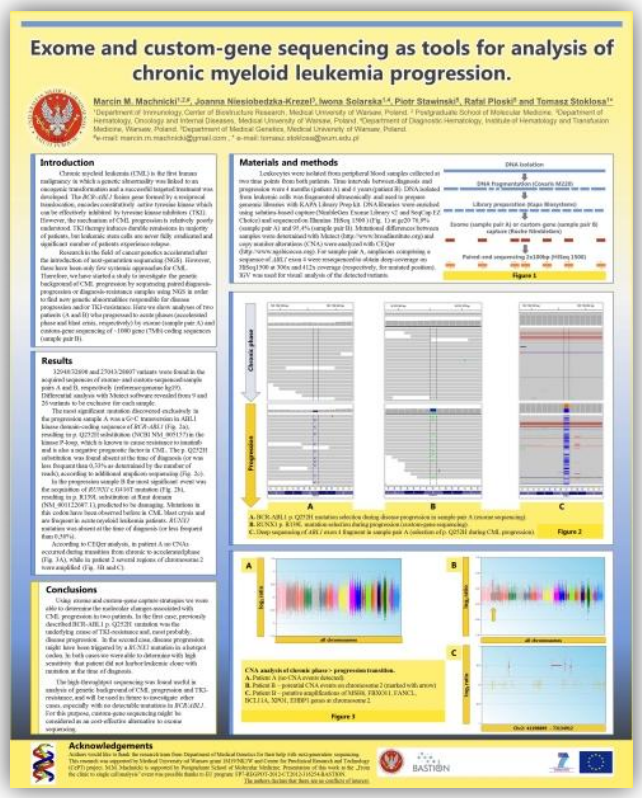
Report on active participation in "NGS Milan 2015: From the clinic To single cell analysis"conference

9 – 10 March 2015 Marcin Machnicki

The "NGS Milan 2015" conference took place at Istituto Nazionale di Genetica Molecolare in Milan, Italy. It was one of the series of meetings organized by bioteXcel consulting company, covering various next-generation sequencing issues, such as DNAseq, RNAseq, single-cell analyses or next NGS quality control. The aim of those meetings is not only to provide participants with newest findings in the NGS field, but also to stimulate networking and knowledge dissemination among them. BioteXcel organized several such meeting in 2014 and 2015.

Topics raised during the meeting in Milan included long non-coding RNAs identification, sequencing circulating tumor cells, identification of causative variants in rare human disorders, integrations of omics data and others.

I had a pleasure to present a poster during the conference:



Title: "Exome and custom-gene sequencing as tools for analysis of chronic myeloid leukemia progression "

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In the poster we presented initial results of our research project focused on genetic changes associated with chronic myeloid leukemia progression. We have shown the results from exome- and custom gene-sequencing approaches in search for genetic events occurring during transition to advanced phases of the disease.

We have found that *BCR-ABL1* kinase domain mutation can be acquired very rapidly and cause progression of the disease even if no other point mutations or copy number alterations are acquired concurrently. Alternatively, a *RUNX1* mutation might be connected with CML progression as we have shown for the second analyzed case.