2. Abstract

Our diploma work involves the introduction of a new method called *Droplet Digital PCR* for the diagnosis and monitoring of the *V617F* mutation of the *JAK2* gene in myeloproliferative neoplasms/diseases (*MPN*/*MPD*). It is similar to the PCR but allows an accurate and absolute quantification of the target. Currently, Real-time PCR (*RQ-PCR*) is weekly used for this mutation detection but the result is not quantified. The introduction of *ddPCR* would allow the quantification of the *V617F JAK2* mutation in order to assess the stage of the disease at diagnosis and to follow the residual disease under treatment.

During our work, we tested different parameters on several kinds of samples (external quality controls, patients and healthy people). We were able to test the sensitivity, specificity, accuracy and cost of the method in comparison with *RQ-PCR*.

From our experiments, *ddPCR* is indeed a good method for quantifying a *rare mutation* and can be used for diagnosis of JAK2 V617F. It has the advantage, in addition to quantification, of being slightly more sensitive and specific compared to the current method. However, it is far from perfect. The experiment requires more reagents and more manipulations, which make it more tedious to perform. The interpretation of the results takes more time as well. The analysis performed in *ddPCR* is also more expensive because *JAK2* is not quantified in *RQ-PCR* by the laboratory. As the method is quite recent, reagents costs are decreasing and make *ddPCR* more attractive.

Keywords: ddPCR, JAK2, V617F, MPN, MPD, RQ-PCR, rare mutation