Childhood acute leukaemias are a heterogeneous group of malignant diseases. Based on cell origin, clinical manifestations, and molecular/chromosomal changes, we distinguish two main subtypes: acute myeloid leukaemia and acute lymphoblastic leukaemia.
Acute lymphoblastic leukaemia (ALL) is the most frequent form of childhood leukaemia. Acute myeloid leukaemia (AML) is predominantly found in adults, being rarer in childhood. In the Czech Republic, the ALL is in childhood diagnosed approximately five times more often compared to AML.
Despite the intensive research, aetiology of leukaemia has not been entirely clarified. So far, we only have knowledge of certain risk factors (ionising radiation, some chemicals and viruses) but in the vast majority of cases the aetiopathogenesis has not yet been made clear.
Some of the answers may be provided by studies dealing with the presence of (pre)-leukaemic cells in a material archived prior to the clinical onset of the disease. Such are for example the so-called Guthrie cards, the dried blood samples collected immediately after birth and used in screening of the newborns for metabolic disorders. The better availability of material collected before the diagnosis of a secondary leukaemia (originally meant for the follow-up of the primary malignancy) might help us in better understanding of the kinetics of pre-leukaemic and leukaemic clones.
In the first part of the thesis, we have used Guthrie cards to retrospectively analyse a large group of AML patients for the presence of their leukaemia-specific markers at birth in a process known as leukaemia backtracking. In the second part, we have backtracked in detail a secondary ALL characterised by the MLL/FOXO3A fusion gene.