Mutation, responsible for genetic diseases, can be inherited from our parents or can arise as a new spontaneous mutation and is called *de novo* mutation. *De novo* mutations can occur in germ cells of parent (germline mutation) or after fertilization (post – zygotic mutation). Post – zygotic type of *de novo* mutations is very interesting from clinical point of view. Carrier of such mutation has two versions of his genome because such mutation is not present in every cell of his body and is called mosaicism. The most significant is tissue or organ where such mutation will arise and it would have an impact for developing the particular symptoms.

At the beginning of embryonic stage, we can distinguish three groups (germ layers) of cells which are precursors of particular tissues and organs: mesoderm – e.g. blood; ectoderm – e.g. nervous system and hair roots, endoderm – e.g. bladder. For many routine biochemical tests, also genetic tests to extract genetic material (DNA), blood is commonly used as a noninvasive and representative material.

The aim of proposed study is to search for post-zygotic mutations in a group of undiagnosed children with neurological disorders e.g. epilepsy, encephalopathy directly in material which reflects nervous system (brain) so in DNA of ectodermal origin. We have selected 8 families where children affect neurological disorders. We want to investigate all coding fragments (so called exome) in DNA from blood of children and their parents to search for mutations present only in DNA of patient – germline mutations. In families in whom we will not detect candidate mutation we will compare exome from child's blood and hair roots to search for post – zygotic mutations.

We believe that such combine study pattern will allow us to detect cause of the disease in almost every included family. We hope to give also reliable genetic counseling and maybe even detect novel mutations responsible for pediatric neurological disorders. We also want to investigate if testing of the specific germ layer, in this case this one reflecting nervous system, can replace blood as a routine material.