Despite significant advances in molecular genetics, which in recent decades have established a large number of genes responsible for the development Macrocephaly but pathogenesis of this condition is not fully known. In most cases, the differential diagnosis Macrocephaly promotes clinical examination of the patient and careful studing of anamnesis.

The clinical and neurological examination helps to suspect a significant number of genetic diseases involving Macrocephaly, including phakomatosis, fragile X-chromosome syndrome, accompanied macrosomia. Neuroimaging methods, including MRI of the brain may help eliminate metabolic diseases, tumors, congenital malformations and hydrocephalus.

Proper differential diagnosis Macrocephaly extremely important, because it allows will start adequate treatment and rehabilitation. It should also be remembered that this condition can be regarded as a risk factor for autism spectrum disorder, especially in the presence of structural brain changes on MRI or family history (presence siblings suffering from (ASD). In this publication we have done attempted to briefly analyze the main reasons Macrocephaly children.