

Metabolism

Metabolism is the chemical process occurring in our bodies, a process in which enzymes, or different forms of protein, play a vital part. Enzymes break down the food we consume and turn it into energy for us to use. Sometimes, metabolism goes wrong because of defective genes in enzymes, passed on by parents.

As a result, toxic substances build up, hindering the work of the enzymes which in turn affects our body functions. This disorder can damage different organs in our bodies. If the nervous system is affected, the sufferer comes down with what is known as a neuro-metabolic disease.

Neuro-metabolic diseases can cause mental retardation, epilepsy, movement disorders, muscle weakness and visual and hearing impairment. To diagnose such disorders, doctors need to carry out clinical examinations, biochemical investigations, such as blood, urine and cerebrospinal fluid examinations and other such tests. As these disorders are caused by genetic mutation, DNA tests may confirm the diagnosis.

Some of these disorders can be controlled effectively through dieting and medications. Early diagnosis and life-long treatment, in some cases, can prevent severe damage to the brain. Unfortunately, treatment options for some progressive and untreatable disorders are limited to dealing only with the symptoms.