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Sabine Scholl-Bürgi, MD

*"*KDT for inborn disorders of metabolism*"*

ABSTRACT

Inborn disorders of metabolism (IDM) belong to the rare diseases. They can be caused by defects of an enzyme, a transporter or a co-factor. To date, more than 1450 different IDMs are known.

Therapeutic options exist for some of these disorders, such as nutritional therapies for phenylketonuria or other aminoazidopathies, or enzyme replacement therapies for lysosomal storage disorders. KDT are also increasingly used in IDM.

In principle, two mechanisms of action can be distinguished: on the one hand, KDT can directly influence the pathophysiology of the underlying IDM as is the case with GLUT1-DS or pyruvate dehydrogenase deficiency. On the other hand, KDT is used to treat symptoms caused by IDM (mostly cerebral seizures).

In order to avoid causing side effects by altering the metabolism in the course of KDT, the underlying pathophysiology of IDM should be known and, if necessary, the therapy target should be adjusted accordingly. For this reason, KDT often needs to be individualized.