Next generation sequencing. New additions to molecular assessment and diagnostics of metabolic defects

Abstract:
The introduction of next generation sequencing is revolutionizing not only molecular genetics and different fields of research but also the diagnostics of genetic diseases. The new sequencing methods facilitate a simultaneous and cost-efficient analysis of thousands of genes which allows a genome-wide search for disease mutations in research and diagnostic settings. The identification of new genes and gene variants discloses new fields in research. Furthermore methods such as exome sequencing will largely replace established diagnostic methods in the future. It is to be expected that automated, cost-saving and time-saving sequencing methods will simplify and accelerate diagnosis and treatment of patients not only with metabolic but also with other kinds of diseases.
TUM Einrichtung:
  r Humangenetik

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