4 The Unique Advantages of Metabolomics in Disease Identification

4.1 Common metabolomics technology platforms

Metabolic research mainly employs two techniques: mass spectrometry and nuclear magnetic resonance. Mass spectrometry is often used in conjunction with chromatography, and it is highly sensitive, capable of detecting subtle changes in metabolites, which is particularly important for neuro-metabolic diseases as the biochemical changes in these diseases are usually very minor. Liquid chromatography-mass spectrometry is the most commonly used in clinical research, with a wide range of applications, and it can analyze samples such as blood and cerebrospinal fluid (Smusz et al., 2025). Gas chromatography-mass spectrometry has an advantage in separating organic acids, and organic acids are key detection indicators for many congenital metabolic problems (Gątarek and Kałużna-Czaplińska, 2025).

Although the sensitivity of nuclear magnetic resonance is lower than that of mass spectrometry, it is highly accurate in quantification, has good repeatability, and the sample processing is simple, making it suitable for long-term tracking studies. With technological advancements, both methods can detect more metabolites related to the nervous system. Some mass spectrometry platforms specifically designed for rare neuro-metabolic diseases can stably detect hundreds of related metabolites and have begun to be used in clinical testing (Smusz et al., 2025).

4.2 Discovery and validation of disease-related metabolic markers

Metabolomics can reflect the changes brought about by the combined action of genes and the environment. This can help identify the metabolic characteristics related to diseases and provide directions for disease diagnosis and research. Taking Parkinson's disease as an example, many studies have shown that the metabolic substances such as fatty acids and amino acids in patients' bodies have changed. These changes not only distinguish patients from healthy individuals but also indicate possible abnormalities in mitochondrial function. In multiple sclerosis and muscular dystrophy, abnormal energy and lipid metabolism have also been found, providing references for finding disease markers and treatment methods (Wijekoon et al., 2025; Smusz et al., 2025).

The advantage of metabolomics is that it first conducts a broad screening and then focuses on targeted verification. Although this field is still in its early stage of development, some of the identified biomarker combinations can be repeatedly verified in different populations and platforms. In the future, they are expected to be used in clinical treatment. In some rare neuro-metabolic diseases, using high-resolution mass spectrometry to analyze cerebrospinal fluid not only proved the reliability of the known biomarkers but also determined the normal range related to age, laying the foundation for finding new biomarkers (Smusz et al., 2025). In summary, the relationship between metabolites and diseases is gradually becoming clearer, and the role of metabolomics in the development of clinical biomarkers is becoming increasingly important.

4.3 The application value of metabolic monitoring in efficacy evaluation

The sensitivity of metabolic changes is very high, enabling them to promptly reflect the body's condition. This makes metabolomics suitable for assessing treatment efficacy and changes in the condition. In the research on Parkinson's disease, long-term observation revealed that the use of levodopa treatment would alter the metabolic state in patients' plasma, adjust the metabolism of some amino acids, and to a certain extent, improve the imbalance of bile acids. These metabolic changes are more accurate than clinical scores and can better reveal the therapeutic effect of the drugs. In the research on multiple sclerosis, metabolomics also found that after treatment with different drugs, lipid metabolism changed, indicating that some metabolites may become indicators for judging the treatment effect or the condition (Smusz et al., 2025). Incorporating metabolic monitoring into treatment decisions helps predict drug responses, detect neurotoxicity early, and provide references for personalized medication.

For neurodegenerative diseases such as Alzheimer's disease and Parkinson's disease, conventional clinical and imaging indicators often fail to sensitively reflect the early neurological changes. Therefore, metabolomics is highly anticipated. By using high-resolution mass spectrometry and nuclear magnetic resonance technology, only a small amount of blood or cerebrospinal fluid sample is needed to simultaneously determine hundreds of