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EDITORIAL

Multi-omic synergy and clinical diagnosis: toward precision medicine and early detection

Sinergia multiómica y diagnóstico clínico: hacia una medicina de precisión y detección temprana

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For decades, traditional clinical tests (complete blood counts, blood glucose, lipid profile, urinalysis, or
diagnostic imaging such as X-rays) have been essential for disease diagnosis; however, these often detect physiological or anatomical changes at relatively advanced stages of illness. Today, technological advances allow us to speak of the "omics sciences," disciplines that provide a more comprehensive understanding of molecules which, collectively, constitute the mechanisms governing the biological functions of organisms. When integrated with traditional clinical tests and family history, they hold promise for improving early detection, prediction, and diagnosis of diseases [1,2].

Genomics in disease diagnosis and prediction

Among the omics sciences that may play a particularly important role in optimizing disease diagnosis is genomics, which enables the identification of inherited genetic variants or somatic mutations associated with the presence or risk of developing a disease. For example, mutations in the BRCA1 and BRCA2 genes are associated with an increased risk of breast and ovarian cancer; their timely identification would allow for monitoring and disease prevention [3,4]. In this way, genomics complements traditional mammography or breast ultrasound by stratifying patient risk, and if the tumor has already developed, somatic genomics through sequencing can reveal genetic alterations that confirm the diagnosis and classify subtypes beyond what the microscope shows, thereby guiding personalized therapies.

Type 2 diabetes (T2D) can be predicted using clinical factors, but genomics can also improve risk prediction. T2D has a complex genetic basis, so identifying associated genetic variants is necessary for better understanding and prevention of the disease. Some meta-analyses in different populations have identified susceptibility loci such as SSR1-RREB1, associated with fasting glucose regulation; POU5F1-TCF19, associated with fasting insulin regulation and insulin resistance; and ARL15, associated with the major histocompatibility complex, which is also essential in the immune response [6].

Familial hypercholesterolemia (FH) is another genetic disorder, associated with elevated cholesterol levels and premature cardiovascular disease. In patients with suspected FH and low-density lipoprotein (LDL) levels above 190 mg/dL, genetic testing of the LDLR, APOB, or PCSK9 genes can confirm the diagnosis and distinguish FH from other causes of dyslipidemia. It has been observed that incorporating genomics increases the number of confirmed FH diagnoses compared with detection based solely on clinical and analytical criteria [6,7].

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Transcriptomics as a complementary diagnostic tool

Transcriptomics studies ribonucleic acid (RNA) as the set of transcripts expressed in a tissue or cell, providing insight into gene expression patterns. For example, in breast cancer, the expression of certain genes in tumor tissue is analyzed to estimate recurrence risk and chemotherapy effects. Combined with traditional factors such as tumor size, histological grade, nodal status, and hormone receptor status, this information allows for more precise therapeutic decisions [1,8]. Furthermore, transcriptomics enables early, non-invasive detection through the analysis of RNA in peripheral blood, or "liquid biopsy," detecting circulating tumor RNA molecules including microRNAs, iRNAs, and snoRNAs. Changes in their levels act as early biomarkers of several common cancers (breast, colon, lung, liver, pancreas, kidney, etc.) or chronic infectious diseases [1,9].

Proteomics and clinical biomarkers

Proteomics enables large-scale measurement of proteins present in a biological sample. Since many traditional clinical tests already measure individual proteins (e.g., insulin, troponin, hormones), proteomics expands the scope by discovering new markers and protein combinations that enhance disease detection. Tumors also release proteins as a defense mechanism against the host's immune response; these can be detected in blood before the tumor becomes visible through radiological imaging. Today, proteomics allows for the development of panels that identify multiple proteins, which, when integrated with circulating DNA analysis, can detect different types of cancer at early stages with a simple blood test [1].

Regarding diabetes diagnosis, proteomics may help detect diabetic nephropathy, not only through the traditional detection of microalbuminuria (minimal albumin excretion in urine), but also by identifying collagen fragments and other proteins that rise years before urinary albumin increases. This information would allow a nephrologist to recommend improved glycemic and blood pressure control before clinically evident renal deterioration occurs [10].

Metabolomics in the early detection of metabolic alterations

Metabolomics consists of the broad analysis of metabolites (sugars, amino acids, fatty acids, etc.), which reflects the overall metabolic state of an organism and provides enormous complementary value to classical clinical tests, particularly for detecting subtle metabolic imbalances that precede evident clinical manifestations. An example is type 2 diabetes (T2D) and other related metabolic disorders, in which specific plasma amino acid signatures are altered years before the onset of hyperglycemia, when glucose or HbA1c levels may still be within the normal range [11].

In cancer, aggressive tumors often consume high concentrations of glucose and produce abnormal metabolites; therefore, metabolomic analysis of fluids can complement imaging in early detection by identifying such subtle changes [1].

In the field of non-oncological metabolic diseases, such as non-alcoholic fatty liver disease, metabolomic profiles of fatty acids and other blood metabolites have been identified, which correlate with hepatic inflammation and improve the identification of at-risk patients beyond traditional liver tests [12].

Lipidomics and advanced lipid profiling

Traditional lipid analyses in clinical practice, such as total cholesterol, LDL, HDL, and triglycerides, provide a limited view that can be enriched by lipidomics, which quantifies different lipid species (phospholipids, sphingolipids such as ceramides, fatty acid subtypes, etc.) that may play crucial roles in cardiovascular and metabolic diseases.

A "lipidomic risk score" has been developed, incorporating specific ceramide concentrations in blood along with traditional lipid markers to estimate the risk of myocardial infarction or stroke, which may not be detected by LDL cholesterol measurement alone. Furthermore, adding these emerging lipidomic markers to traditional cardiovascular risk models (including cholesterol, blood pressure, smoking, etc.) significantly improves patient stratification [11]. Likewise, in diabetes and prediabetes, studies have shown that long before fasting glucose levels rise, changes in certain plasma phospholipids and chain-specific triglycerides can already be detected in individuals who later developed diabetes, suggesting that lipidomic analysis could improve the ability to predict disease onset [12].

Bioinformatics integrating omics sciences and clinical data in precision medicine

Within this framework, bioinformatics occupies a central place in precision medicine by processing and

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merging large amounts of data from various omics sciences with traditional information, such as electronic medical records, laboratory analyses, and imaging, in order to propose personalized treatments [13]. Moreover, integrating these heterogeneous data with machine learning algorithms and neural networks facilitates the extraction of complex patterns. For example, artificial intelligence platforms have been designed to correlate clinical and genetic profiles of patients with autoimmune diseases, incorporating diagnostic imaging (X-rays, CT scans, MRI, digital pathology) along with genomic and clinical data, thereby enabling risk classification and optimization of personalized therapeutic strategies [13,14].

Concrete examples include AI algorithms applied to well-characterized cohorts to predict disease onset or patient prognosis, as in the context of COVID-19. The use of SARS-CoV-2 genomic sequencing combined with multi-omic analyses (immunomics, proteomics, metabolomics) has enabled prediction of disease severity and stratification of patients by risk of adverse outcomes. A similar approach has been applied in prediabetes and T2D, combining metabolomic and clinical history data to predict progression [15,16].

Medicine–biology synergy

Perspectives from biology and medicine converge in the field of translational medicine, making close dialogue between the two essential: the physician, who cares for and diagnoses the patient, raises unmet needs, while the scientist, who seeks to understand disease at a more fundamental level, provides solutions based on omics exploration [2]. Imagine a scenario where a single marker could detect a given disease or distinguish subtypes that appear identical on imaging, while on the other side a proteomic signature is identified that fulfills that role.

In conclusion, the omics sciences provide a deep and personalized view that complements the broader picture offered by traditional analyses, enabling earlier and more accurate diagnosis, enriching disease understanding, and ultimately improving clinical outcomes (see Figure 1). Multidisciplinary collaboration plays a fundamental role in this process, spanning from clinical approaches to the adoption of innovative diagnostic tools, including the integration of multi-omic data with traditional clinical evaluation, which will become increasingly common as omics technologies continue to advance in precision and accessibility.

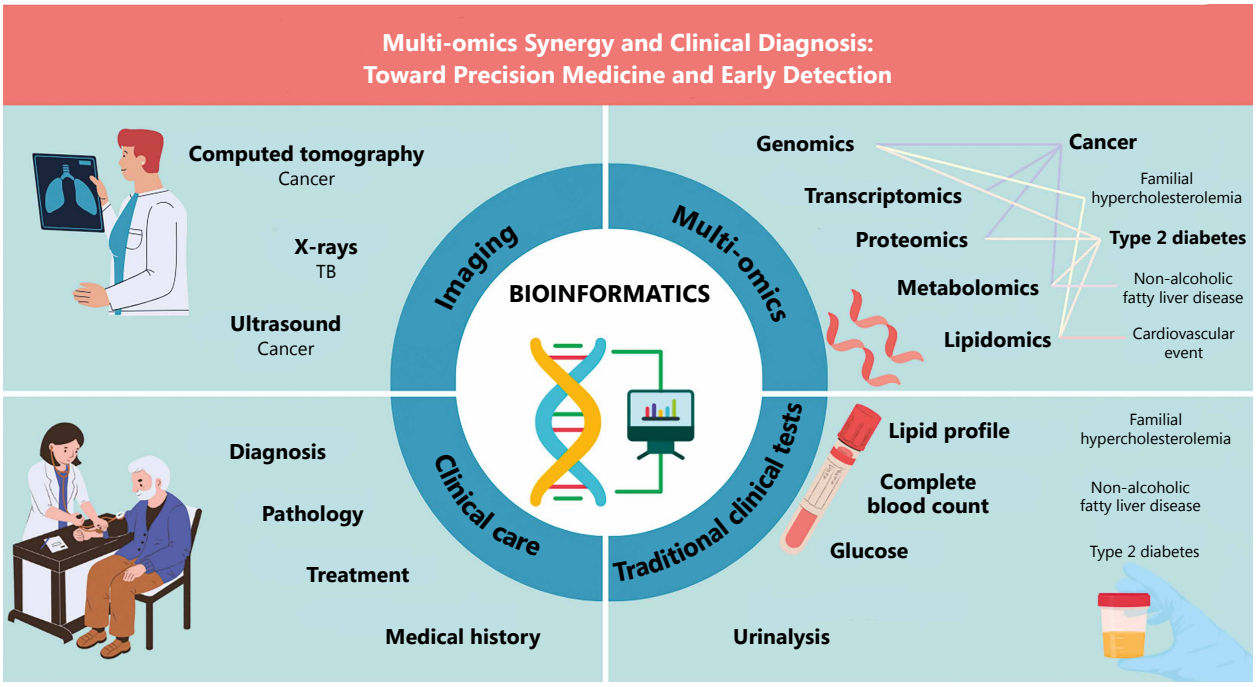

Figure 1. Multi-omics synergy

* Multi-omics analysis is the convergence of clinical data, laboratory data, radiological images, and the results of bioinformatic analyses of DNA, proteins, and metabolites present in blood, urine, or any body fluid.

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Conflict of interest statement

The authors declare no conflicts of interest.