NGS and AI in prenatal diagnosis: Issues and ethical implications

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The integration of Next-Generation Sequencing (NGS) into prenatal diagnostics offers unprecedented genomic insight, yet its marriage with automated AI interpretation of sequencing data from fetal DNA (either cell-free in the maternal circulation or extracted from fetal cells after amniocentesis or chorionic villi biopsy) introduces a fraught ethical landscape. A primary concern is the "black box" nature of these algorithms; when clinicians cannot parse the logic behind a variant's classification, the foundation of informed consent begins to crumble. This lack of transparency doesn't just hinder shared decision-making—it risks automating historical biases, potentially widening existing gaps in health equity.

Furthermore, the clinical stakes of misinterpretation are uniquely high in a prenatal context. As the International Society for Prenatal Diagnosis (ISPD) has noted, fetal sequencing is notoriously complex due to the ambiguity of genotype-phenotype correlations. Without a "human-in-the-loop" to provide expert context, automated systems may mislabel uncertain variants, leading to life-altering clinical decisions based on incomplete or misunderstood data. This risk extends to the accidental discovery of sensitive information, such as non-paternity or adult-onset conditions. Without robust, multidisciplinary oversight to filter these outputs, we risk violating parental privacy and undermining the very trust that underpins maternal-fetal medicine.