

Approach to Genetic Counseling in Resource-Limited Settings: A Case-Based Analysis from an Obstetric Outpatient Cohort

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Objective

To analyze the presentation of couples to the outpatient department, with particular focus on the status of the affected proband, the presence or absence of a confirmed genetic diagnosis, and the clinical approach adopted when diagnostic information is unavailable, incomplete, or potentially inaccurate—scenarios frequently encountered in middle- to low-income countries. The study also aims to propose a pragmatic and resource-sensitive framework for the management of ongoing pregnancies or for planning future pregnancies in such settings.

Methods

To retrospectively review case records of couples seen in the outpatient department from February 2021 to January 2025, evaluating the status of the affected proband, presence of a confirmed genetic diagnosis, and the management strategy in the absence of prior diagnostic data.

Results

This study reviewed four distinct clinical scenarios frequently encountered in genetic counseling within resource-limited settings: Couples with affected offspring and a confirmed genetic diagnosis (n=6): These cases allowed for straightforward application of targeted prenatal diagnostic strategies in subsequent pregnancies. Couples with affected offspring and a strong clinical suspicion of a genetic disorder but no established diagnosis (n=4): These cases posed interpretative challenges and required re-evaluation of phenotypic details and a stepwise plan for genetic workup prior to conception or early in pregnancy. Couples with deceased offspring or intrauterine demise without available genetic samples (n=3): These scenarios highlighted the critical limitations in postmortem genetic evaluation and emphasized the importance of counseling about recurrence risk using syndromic clues, family history, and imaging archives. Couples with all offspring affected and incomplete or misguided genetic workup (n=2): These cases underscored the consequences of fragmented diagnostic efforts and reinforced the need for centralized, multidisciplinary review. Each scenario demonstrated the necessity of a tailored, context-sensitive approach—particularly in middle- to low-income countries where prior investigations may be lacking, incomplete, or inaccurately interpreted. Such individualized strategies are essential for meaningful interpretation of genetic findings and for optimizing reproductive decision-making, including the planning of future pregnancies and prenatal diagnostic pathways.

Conclusion

A strong grasp of genetics is essential for obstetricians, who often act as the first point of contact for couples with a history of affected offspring—especially in low- to middle-income settings where access to specialized genetic services is limited. This study highlights the varied and complex real-life scenarios that require obstetricians to interpret incomplete or absent genetic information and guide reproductive decisions. Enhancing practical genetic literacy and promoting case-based, context-sensitive approaches are crucial for optimizing prenatal care and outcomes.