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Journal of Down Syndrome & Chromosome Abnormalities

Perspective

Genetic Abnormalities and their Role in Pregnancy Loss: Implications for Diagnosis and Management

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DESCRIPTION

Genetic abnormalities, whether in the conceptus or the parents, can contribute to both sporadic and Recurrent Pregnancy Loss (RPL). In the conceptus, common genetic issues include aneuploidy, copy number variations, skewed X-inactivation and single-gene mutations or disorders. Among parents experiencing RPL, the most well-studied genetic factor is the presence of balanced chromosomal translocations. When evaluating genetic causes in cases of pregnancy loss, chromosomal microarray is often preferred over traditional karyotyping because it provides more reliable results, especially when cell culture fails. However, for parental genetic testing, karyotyping remains the gold standard, as microarray technology may not detect balanced translocations. In cases where a genetic abnormality is identified, Preimplantation Genetic Testing (PGT) has been proposed as a strategy to improve live birth rates, though there is currently insufficient evidence to support its widespread use. In conclusion, while various genetic causes of RPL are recognized, the implications for clinical management remain uncertain when a genetic cause is identified.

Pregnancy loss is a common occurrence, affecting approximately 30% of conceptions and 10% of clinically recognized pregnancies. The genetic causes of both types of losses are significantly similar, even though the majority are sporadic rather than recurring. Depending on when the loss occurs, the underlying reasons of pregnancy loss can change, with hereditary factors being more likely to play a role in early pregnancy. Therefore, it is crucial to categorize losses by their developmental stage rather than just gestational age. Losses can occur at different stages: Preimplantation, pre-embryonic (post-implantation but before the embryo is visible on ultrasound), embryonic (embryo visible on ultrasound, <10 weeks), early fetal (10 weeks-13 weeks), late fetal (14 weeks-19 weeks) or stillbirth (≥ 20 weeks). Accurately documenting the developmental stage is important, as there may be a significant delay between fetal demise and the onset of symptoms or diagnosis.

Many clinicians decided not to use genetic testing following pregnancy loss, as its benefits are not immediately clear. The process is often seen as costly, with limited perceived value for future pregnancy planning and unlikely to alleviate the emotional impact on the couple. Consequently, most professional societies do not recommend genetic testing for sporadic early losses. However, there is greater support for genetic testing in cases of recurrent early losses and sporadic late losses. Identifying a genetic cause in these cases can provide valuable information about recurrence risk and help guide further evaluations, potentially avoiding unnecessary treatments.

CONCLUSION

In conclusion, genetic abnormalities, both in the conceptus and parents, play a significant role in RPL. While chromosomal microarray is a valuable tool for evaluating pregnancy loss, karyotyping remains the standard for assessing parental genetic factors like balanced translocations. Preimplantation genetic testing holds promise for improving live birth rates in cases of identified genetic abnormalities, but more research is needed to validate its effectiveness. Overall, while genetic causes of RPL are well-established, the clinical management of affected couples remains uncertain and requires further investigation. In some instances, genetic abnormalities can influence management strategies and future advances may offer opportunities for treatment or prevention.

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Received: 19-Aug-2024, Manuscript No. JDSCA-24-35836; **Editor assigned:** 21-Aug-2024, PreQC No. JDSCA-24-35836 (PQ); **Reviewed:** 04-Sep-2024, QC No. JDSCA-24-35836; **Revised:** 11-Sep-2024, Manuscript No. JDSCA-24-35836 (R); **Published:** 19-Sep-2024, DOI: 10.35248/2472-1115.24.10.264

Citation: Medi C (2024). Genetic Abnormalities and their Role in Pregnancy Loss: Implications for Diagnosis and Management. J Down Syndr Chr Abnorm.10:264.