Prenatal diagnosis of primary immunodeficiency: an emerging experience in Egypt

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Objective

To present the current status of genetic counselling services and results from one of the largest primary immunodeficiency centers in Egypt. We also intend to show the impact of prenatal diagnosis on the families and the management plan of the diagnosed cases.

Methods

Based on molecular diagnosis of the index cases, prenatal diagnosis was offered to 10 families in 12 subsequent pregnancies. Five different genes were sequenced based on the condition and diagnosis of the index cases.

Results

Six fetuses were either normal or carriers of mutated genes and families decided to continue with the pregnancy. In five pregnancies molecular investigations following CVS proved the fetuses to be affected and HLA typing was performed seeking a suitable related donor for SCT.

Conclusion

In spite of the genetic heterogeneity behind PIDs, genetic counselling should play a critical role in the management and future decisions of the affected families.