

## عنوان مقاله:

Genetic Consultation, Genomics Techniques and further in-vitro fertilization Strategies in families with a child involved with auto-immune hemolytic disorders

## محل انتشار:

هشتمین سمینار کشوری مشاوره ژنتیک و نقش آن در پیشگیری از معلولیت ها (سال: 1398)

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## خلاصه مقاله:

Background and Aim : Auto-immune hematologic disorders in infants include a vast spectrum of complicated and heterogeneous entities involving genetic and environmental elements which sometimes too difficult to discriminate and manage especially in planning for the next pregnancies, unless through a series of molecular genomics experiments like next generation sequencing techniques. In this study we elucidate the relevance of para-clinic data with genetic consultation findings to re-consider the in-vitro interventional strategies for the parents in their plan for further pregnancies to prevent repeated cases of involved child. Methods : In our study, NGS technology, efficiently contribute to study the genetic mutations of complicated auto-immune infantile hemolytic anemia in a group of 7 referred cases during a period of 12 month. Each patient considered for detailed genetic consultation interview succeeding by appropriate mutation determination techniques. Results : Two out of seven patients has been detected as homozygote cases for the pathogen mutations and the rest were considered as heterozygote, hence there were guided proportionately in planning the for further pregnancies. Conclusion : In-vitro fertilization technologies include all forms of interventions with handling of germ cells and/or zygotes and embryos and are most useful in guiding the families with previously involved child, either in hetero or homozygote forms, although should be selected very carefully to end in success with lowest charges. In our study one may consider an optimal set of strategies like egg or embryo donation as the best choices and recommendations in further pregnancies for each family proportionately by combination of genetic consultation skills and genomic study techniques.

## کلمات کلیدی:

genetic consultation, Genomic study, in vitro fertility

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