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LOOKING INTO THE CHALLENGES IN EXPANDED CARRIER SCREENING

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Introduction: Carrier screening can be defined as genetic investigation of carrier status of a person or a couple for a disorder with recessive inheritance. Expanded carrier screening allows pan-ethnic testing of all individuals. The major goal of carrier screening is providing more reproductive choices for a carrier couple. Screening programs should have important criteria such as voluntary participation, pre- and post-test counselling, selecting a test with known sensitivity, specificity and predictive values.

Methods: Data were obtained from 296 preformed carriers screening for 600 autosomal recessive and X-linked recessive disorders in Iranian population. This investigation was performed by Next Generation Sequencing (NGS), Gap-PCR, long PCR, qPCR and PCR-Sanger sequencing.

Results: This study revealed that 78% of screened individuals have at least one previously reported variant or novel variant with high putative effects. From 388 detected variants, 276 variants have been previously reported as pathogenic variants. Detailed carrier frequencies for different disorders in the study, and associated genes and mutations will be presented in the meeting. Furthermore, this study uncovered many other challenges that are being faced in carrier screening such as dealing with novel variants, consanguineous couples with carrier status for multiple disorders, requests of prenatal diagnosis for suspected pathogenic variants and so on.

Conclusion: The introduction of Next Generation Sequencing (NGS), provides more opportunities for performing carrier screening for multiple autosomal and X-linked recessive disorders. A comprehensive study is necessary to address many inevitable challenges the society faced in implementing any large scale carrier screening program.

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