A standard tool to analyze, interpret and report pathological sequence variants related to recessive diseases in NGS data

Background

Although individually uncommon in general populations, Mendelian diseases are collectively reported to account for ~20% of infants mortality and ~10% of pediatric hospitalizations.

More than 2000(Autosomal and X-linked) recessive disorders have been identified.

Prevalence of an offspring affected with a recessive disease is higher among consanguineous couples. Pre conceptual detection of carrier status and genetic counseling enables prevention of disease and providing them with informed reproductive choices. However, the optimum method to conduct pre conception career screening and the interpretation and responsible reporting of the results are vital to prevent misinformation and ethical

Traditional carrier screening methods focus on certain

ethnic/geographical populations with a higher prevalence of certain

Recessive diseases.