1 percent of European couples are at risk of having a child with a severe recessive genetic disorder

Based on 6,447 exome sequences of healthy, genetically unrelated Europeans of two distinct ancestries… researchers calculated that each individual is a carrier of at least two pathogenic variants in currently known autosomal-recessive (AR) genes, and that about 1 percent of European couples are at risk of having a child affected with a severe AR genetic disorder, they wrote in a paper published [March 25] in the American Journal of Human Genetics.

Further, the researchers found that the risk of having an affected child is 16.5-fold higher for first cousins, and is significantly higher for skeletal disorders and intellectual disabilities because of the distinct genetic architecture of those conditions.

"Europeans carry two to four variants on average. This was suspected before, but this is the first time that we actually see data that proves it," co-corresponding authors and Radboud University Medical Center researchers Christian Gilissen and Hans Brunner said in an email.

"More importantly, we now know that 1 percent of non-consanguineous couples are at risk of having children with severe recessive disease. Even though the two populations that we looked at have their own variants, they show striking resemblance in terms of overall carrier frequencies for different disorders."

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