

Genetic disease spotting through genome analysis

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The new genome analysis software tool can spot precise genetic changes that cause disease in more than three billion letters of DNA code that make up the human genome.

This is done by linking to a database of clinical information from individuals with genetic diseases to pinpoint DNA changes that are known to cause illness.

The genome analysis software also predicts the consequences of DNA changes, helping to identify disease causing differences that are not already linked to a known condition. In addition, the software scans databases of genetic information from healthy individuals to rule out DNA differences that look as though they may cause a disease but are harmless - minimising the risk of false diagnoses. Experts say the system is especially useful for diagnosing disorders that are induced by many different genes, such as severe intellectual disabilities in children.

This new system is freely available online, which will help to overcome this bottleneck and make it easier to diagnose genetic conditions in clinical practice and in research programmes.

So, It can be say that this genome analysis software has changed the approach, pace and success of molecular diagnosis of several genetic conditions. Nevertheless, this technology is still under development and proper training of clinical geneticists and genetic counsellors will able to improve this situation.

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