Most advanced genetic map may pinpoint diseases

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People of west African ancestry have hotspots of gene recombination that are not seen in people of European descent. Such hotspots may be linked to genetic errors that contribute to congenital diseases like anaemia, so the finding could help identify the origin of diseases that particularly affect these people.

The finding comes from the most advanced map of the genome to date, which could also help in the study of genetic diseases in those with European ancestry.

Every normal body cell contains two copies of each chromosome, one inherited from each parent. During meiosis – the type of cell division that produces sperm and eggs – corresponding chromosomes from the individual's mother and father exchange fragments of DNA so that the resulting sperm or eggs contain genetic material from both parents. This process is called recombination, and happens at certain "crossover" sites of the genome.

Recombination can go wrong, however – a fragment of genetic code can be accidentally deleted or inserted where it does not belong. Such errors are often linked to genetic diseases.

Mixed ancestry

David Reich of Harvard University and his colleagues devised algorithms to analyse genetic data collected from nearly 30,000 African Americans and identified around 2.1 million crossovers where recombination occurred.

Before Reich's work, the most accurate genetic map was based on data from 15,000 Icelandic parents and their children, which revealed 500,000 crossovers.

Reich's team took advantage of the mixed ancestry of their participants to locate a higher number of crossovers. That's because the average African American has about 80 per cent west African ancestry and 20 per cent European ancestry, producing a genome with long an unbroken segments of either African or European ancestry. Reich's team could pinpoint crossovers by looking for segments of European ancestry that had been punctuated by a fragment of west African DNA, or vice versa.

African hotspots

When the researchers compared their new map with earlier maps that covered solely European genetics, they found important differences at a fine scale – namely about 2500 recombination hotspots that are usually active in people of west African ancestry but nearly always inactive in Europeans.

The 2500 recombination hotspots unique to people of west African ancestry may be associated with genetic diseases found only in such people.

"We found something new about recombination," says Reich. "We had somewhat naively assumed recombination rates are identical across humans. This is a fantastic new resource useful for gene mapping and gene discovery."

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