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RESEARCH NOTE

Using genetic disease data to explore human ancestry: a probe based on data from the population of India

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Long-term studies on human evolution have focussed largely on the fossil record, followed by the analysis of ancient DNA (Reich 2018). Here we show how contemporary genetic diversity can contribute through the major disease databases of online Mendelian inheritance of man (OMIM) and, the Indian genetic disease database (IGDD). We note how similar studies, for Iran and for the aboriginal populations of Australia and New Zealand could contribute. Here we show how a simple genetic probe could be applied to this problem, based on the distribution of dominant and recessive inheritance among genetic diseases.

A simple but important issue bearing on human evolution is whether a genetic disease is inherited in dominant or recessive form. In the Indian and European-American populations, we identify 12 diseases listed in OMIM for the European and American population as exclusively autosomal dominant, but which have predominantly autosomal recessive cases listed in IGDD, the corresponding database for India. We suggested that this finding reflects the population bottleneck of the last ice age, acting on the European but not the Indian populations (Mitchison and Mitchison 2018).

The 12 diseases in question here are chronic pancreatitis, congenital cataract, familial hypercholesterolaemia, Gilbert syndrome, haemophilia A, haemophilia B, hypertrophic cardiomyopathy, isolated growth hormone deficiency type 1, lattice corneal dystrophy, primary congenital glaucoma, spinocerebellar glaucoma, spinocerebellar ataxia and venous thrombosis.

Together these now constitute a probe that could be used to explore the ancestries of other populations of interest, such as the aboriginal populations of Australia and New Zealand, and the present population of Iran, where it could indicate whether they lean towards Indian or European ancestry. Mode of inheritance data for even one or two of this panel of diseases would in each case be indicative and could encourage collection of further data. So also could data for Iran, a south Asian neighbour between India and Europe.

References

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