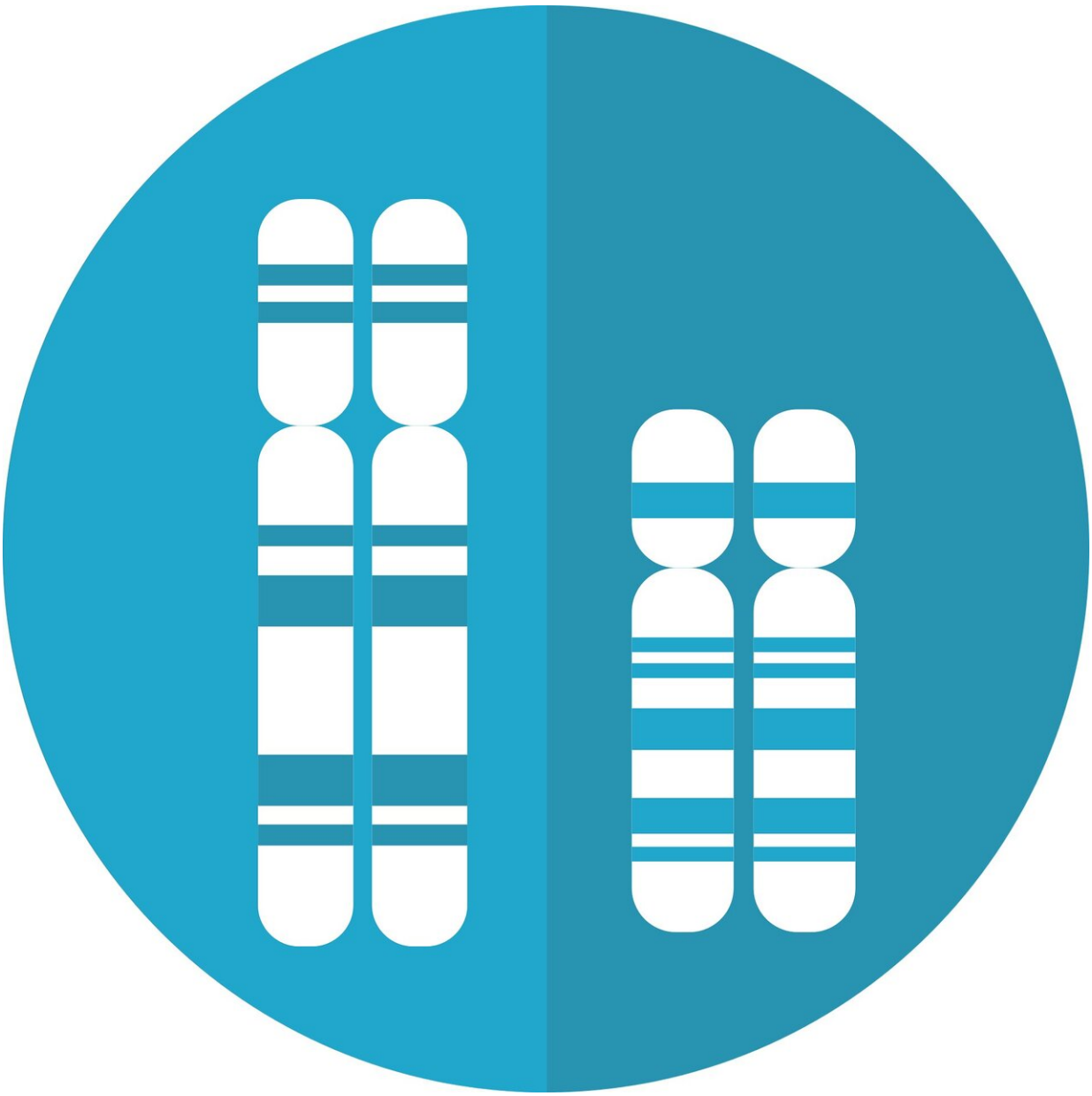
Medical Xpress

Chromosomal changes implicated in disease linked to social and economic disadvantage

September 23 2019



Chromosomal changes implicated in disease are linked to social and economic disadvantage, finds a study of 473 families, published online in the *Journal of Medical Genetics*.

The findings add to the evidence on the biological factors that may be involved in disadvantage, say the researchers.

Chromosomes are the thread-like structures found in the nuclei of all living cells. They carry genetic information in the form of genes.

It has long been thought that genetic conditions may affect the social and economic status of patients and their families, but few studies have tested this theory, say the researchers.

Socioeconomic status—the measure of a person or family's social and economic standing—is based on several factors, including household income, education, and employment. It is strongly implicated in health and disease.

The researchers focused on a particular type of genetic change implicated in disease. The changes, known as pathogenic copy number variants, describe extra or missing sections of genetic material.

They studied the anonymised results of chromosome tests, performed between 2010 and 2017, mostly from children suspected of having a genetic condition. These were taken from a database of more than 17,000 DNA samples at the Manchester Centre for Genomic Medicine.

In all, 473 cases of chromosomal changes implicated in disease were

found, for which information on residential postcode and heritability were also known.

The postcodes were then mapped to the Index of Multiple Deprivation (IMD), an official measure that ranks every area of England by level of deprivation.

This showed that people with these particular changes tended to live in more deprived areas.

When the results were divided into those changes inherited from a parent and those which had arisen spontaneously, they found that people with the inherited changes were more likely to live in areas of even greater deprivation.

This is an observational study, and as such, can't establish cause. But the findings show that even in the absence of a medical diagnosis, chromosomal changes implicated in disease may have a discernible effect on socioeconomic status, suggest the researchers.

"Lower socioeconomic status in families with medically relevant inherited pathogenic and likely pathogenic [genetic changes] with milder phenotype [lower risk of disease] could therefore be due to cumulative multigenerational consequences of these subclinical effects," they write.

More information: Presence of pathogenic copy number variants (CNVs) is correlated with socioeconomic status, *Journal of Medical Genetics*, DOI: 10.1136/jmedgenet-2019-106292

Provided by British Medical Journal