PARENT-of-ORIGIN EFFECTS (imprinting)

Imprinted genes can affect the phenotype depending on the parent from whom the alleles are inherited. That is, identical genetic variations can have different phenotypic effects as a consequence of their parent-of-origin (PofO). Prader-Willi syndrome is probably the best known example in humans. In the clinic, the identification of the phenomenon requires knowledge of the patient's genealogy. The authors of a paper in BioRxiv[1] used a new probabilistic approach to deduce the PofO of individual alleles, in the British biobank, that does not require parental genomes or prior knowledge of genealogy. They examined 59 biomarkers and 38 anthropomorphic phenotypes for PofO effects and found 101 significant associations that contribute to the genetics of complex traits. This study (and similar studies underway with other biobanks) substantially expands the catalogue of imprinting genes in the human population.

[1] https://www.biorxiv.org/content/10.1101/2021.11.03.467079v1