



**Figure 2. Heterodisomy and isodisomy: imprinting or unmasking of a mutant recessive allele?** The two main types of uniparental disomy (UPD) are represented, using maternal UPD7 as an example. (a) Heterodisomy; two different copies of the maternal chromosome 7 (inherited from both maternal grandparents). A recessive mutation (labelled 'X') present in a biallelically expressed gene is masked by the second maternal homologue. (b) Isodisomy; two replica copies of a maternal chromosome 7 homologue. If the mother is a carrier of a recessive mutation on this homologue, it will be 'unmasked' and cause a recessive genetic disease. Imprinting effects are seen in both heterodisomy and isodisomy, but demonstration of complete heterodisomy can rule out aetiology due to mutation of a recessive gene (**fig002gml**).

Heterodisomy and isodisomy: imprinting or unmasking of a mutant recessive allele?