FRAXF syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. FRAXF syndrome. ORPHA:100974

FRAXF syndrome was originally identified in a family with developmental delay and an expanded CCG repeat at the folate-sensitive FRAXF fragile site. Since this initial description, FRAXF has been associated with a range of manifestations but no clear phenotype has been established.