



Rare Diseases: Skeletal disorders - Skeletal dysplasias #2

(1) If no previous genetic testing, advise recruitment as singleton. However, if previous genetic testing was normal then recruit with affected relatives

- (2)
- Short stature (no other syndromic cause)
 - Dentinogenesis Imperfecta
 - Blue sclerae
 - Joint hypermobility
 - Deafness
 - Facial Dysmorphism characteristic of OI / bone fragility
 - Unexplained joint contractures

ADVISED testing:
 AMELX, FAM83H, ENAM, c4orf26, KLK4, MMP20, WDR72, LAMB3, LAMA3, ITGB4, COL17A1, LTBP3, FAM20A, FAM20C, DLX3, STIM1, GPR68, LAMC2, PEX1, PEX6

Secondary and systemic cause, fluorosis

Clinical and radiological diagnosis

Amelogenesis imperfecta

Condition

Osteogenesis Imperfecta (OI)

(1) Clinical diagnosis based on childhood onset bone fragility (>1 fracture and/or low impact fractures) AND ≥1 of (2)

Other syndromal short stature without bone fragility
Secondary causes of bone fragility

ADVISED testing:
COL1A1, COL1A2, c-14C>T IFITM5 testing, OI gene panel

Unexplained skeletal dysplasia

Unknown skeletal dysplasia OR known skeletal phenotype

Disproportionate short stature with non skeletal aetiology
Other non skeletal causes of short stature
ADVISED testing: according to phenotype

Genes (according to local standard practice)
Exclusion Criteria

Inclusion Criteria

Professor Julian Barwell
Dr Corrina Powell
Helen Harrison

University Hospitals of Leicester NHS Trust

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