



A small 15-year-old girl with normal intelligence and motor function presents with an enlarged bulging forehead, protruding jaw and frontal bossing.

?Diagnosis

Radiological features and Genetics

Diagnosis: ACHONDROPLASIA

X RAY FEATURES

Achondroplasia

1. Square iliac bones (“champagne glass”)
2. Fattened acetabulae and short femoral necks.
3. Lateral bowing of the distal humerus.

Craniofacial features:

Shortened base of skull with small foramen magnum, Prognathism
Large skull with frontal bossing and broad mandible

Skeletal features:

Shortening of the proximal limbs
Brachydactyly and trident hand configuration
Limited elbow extension and rotation
Hyperextensibility of most joints, especially the knees.
Genu varum

GENETICS

Point mutation of gene coding FGFR3; Chromosome 4.; AD
Arginine to glycine (nucleotide 1138) in 95%

Normal FGFR: controls growth.

When deficient = increase growth. When mutated = decrease growth.

“Gain of function mutation

Other condition linked with FGFR3: Thanatophoric dysplasia and
Craniosynostosis (FGFR2)

Hypochondroplasia: Tyrosine Kinase domain problem (Normal head)