Syndromic Congenital Muscular Dystrophy

Genetics

-Fukuyama (FCMD; FCMD)

-Muscle‐Eye‐Brain (MEB; POMGNT1)

-Walker‐Warburg (WWS; POMT1/POMT2)

-Congenital Muscular Dystrophy Type 1D (MDC1D; LARGE)

-Mostly AR; but: collagen VI-deficient CMD is AR/AD; LMNA-related CMD is AD (all de novo)

Clinical findings/Dysmorphic features

-Hypotonia and muscle weakness present at birth or during infancy (floppy baby)

-Poor/decreased motor abilities, delay/arrest of motor milestones, joint/spinal deformities

-Onset of manifestations < 2yrs may be a reasonable diagnostic criterion

Etiology

-Prevalence of 1:125,000

Pathogenesis

-Disruption of alpha dystroglycan (integral component of the dystrophin‐glycoprotein complex)

Genetic testing/diagnosis

-Muscle biopsy: dystrophic or myopathic pattern; increased serum creatine kinase; brain MRI: Cobblestone complex (enlarged flat ventricles, flat brainstem, cerebellar hypoplasia)

Others

-LGMD is defined by muscle weakness in late childhood or adulthood