1p36 Deletion Syndrome

Genetics

-Genes unknown; contiguous gene deletion syndrome

-Terminal deletion of 1p36; F:M, 2:1

Clinical findings/Dysmorphic features

-Craniofacial features: straight eyebrows, deeply set eyes, midface retrusion, wide and depressed nasal bridge, long philtrum, pointed chin, large, late-closing anterior fontanel (77%), microbrachycephaly (65%), epicanthal folds (50%), posteriorly rotated, low-set, abnormal ears

-DD/ID of variable degree in 100%; hypotonia in 95%; seizures (44%-58%); structural brain abnormalities (88%); congenital heart defects (71%); eye/vision problems (52%) and hearing loss (47%); skeletal anomalies (41%); brachy/camptodactyly and short feet; abnormalities of the external genitalia (25%); renal abnormalities (22%)

Etiology

-Between 1:5,000 and 1:10,000 births

Pathogenesis

-No genes have been associated with clinical features of 1p36 deletion syndrome

Genetic testing/diagnosis

-Conventional G-banded cytogenetic analysis, FISH, CMA can be used to detect deletions

-Complexity of some deletions may be detected only by CMA.

Others

-Most common terminal deletion syndrome; majority maternally derived