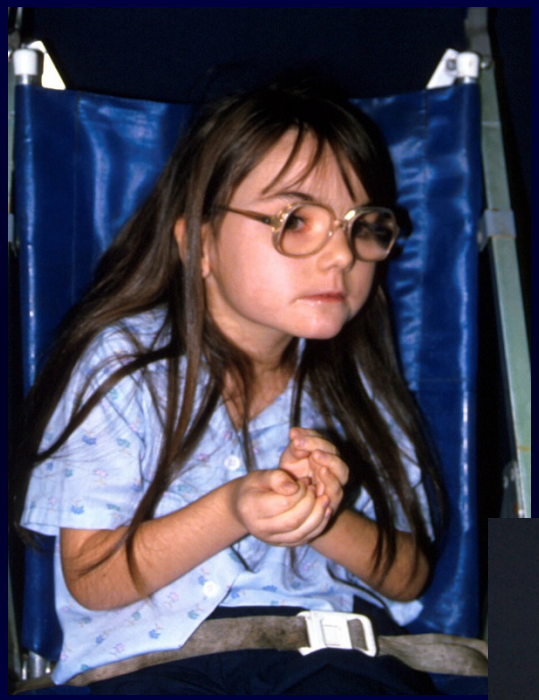
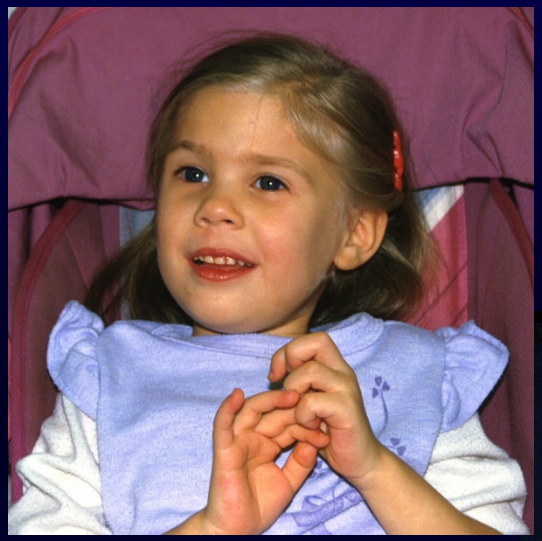
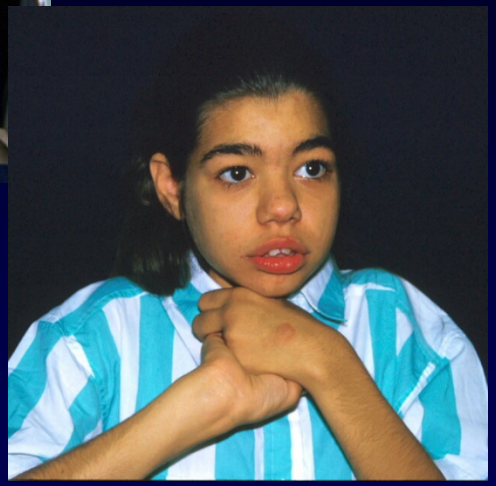
Rett Syndrome

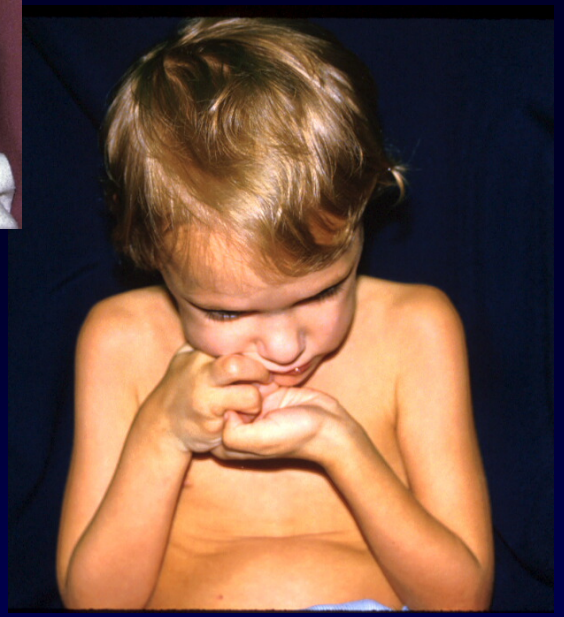
- 1/10,000 incidence
- Almost 100% of cases are female
- Normal family history
- Essentially normal biochemical/CSF/MRI tests
- Gene MeCP2 (Xq28): a methyl binding protein, About 80% have point mutations, 20% have large deletions.

Rett Syndrome









Rett Syndrome: Manifestations

- Baby is normal initially
- Head circumference stagnation begins
- Gait/posture dyspraxia
- Psychomotor regression
- Severe intellectual deficiency
- Loss of purposeful hand movements
- Stereotypical hand movements