FISH ANALYSIS OF CHROMOSOME 15 - PRADER-WILLI SYNDROME

Deletion of 15q11.2-q13

Normal

Deletion
FISH ANALYSIS OF CHROMOSOME 15 - ANGELMAN SYNDROME

Deletion of 15q11.2-q13

Critical Region Probe
Chromosome Reporter Probe

Normal
Deletion
Marfan Syndrome
PIERRE ROBIN SEQUENCE
Beckwith Wiedemann
Microtia
Hemifacial Microsomia
OI Clinical characteristics

- Extreme fragility of bones
- Opalescent teeth (dentinogenesis imperfecta)
- Blue sclera
- Deafness
- Ligamentous laxity
- Skin abnormalities

- Decreased height
- Bowing of limbs
- Scoliosis
- Triangular face
FIGURE 1. A, Young infant with vertebral anomalies, anal atresia, esophageal atresia with T-E fistula, radial aplasia on the right, and thumb hypoplasia on the left. B, Same patient at 2 years of age, with normal intelligence. C, Relative frequencies of some of the other VATER association defects when the patient is ascertainment by virtue of having one of the defects. (From Quan, L., and Smith, D. W.: J. Pediatr., 82:104, 1973, with permission.)

FIGURE 2. Left, Expanded VATER association of defects, including genital anomaly (right).