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Microdeletion Syndromes Detected by FISH – 73 Positive from 374 Cases

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ABSTRACT Fluorescence *in situ* hybridization (FISH) has facilitated the detection of microdeletions seen in Prader-Willi/Angelman (PW/AS), Williams and DiGeorge syndromes. Out of 374 suspected cases tested at Jaslok Hospital in the past 5 years, 73 were positive, including 29 cases of Angelman, 16 of Prader-Willi, 24 of Williams and 4 of DiGeorge syndrome. Male preponderance was seen, mainly in Williams syndrome. The mechanisms causing Prader-Willi and Angelman syndrome include microdeletions, intragenic mutations, uniparental disomy and imprinting defects, though FISH can only detect microdeletions. Metaphase FISH helped to detect 1 case each with deletion of the control (PML) signal and duplication of the critical PW/AS region, which are associated with autism. One suspected case of Prader-Willi syndrome had a Robertsonian translocation t(14;15)(q10;q10) which led to a deletion of a major part of the SNRPN region in 10% cells, resulting in low-grade mosaicism. Another FISH-positive case was due to a reciprocal translocation t(2;15)(q37;q11), where loss of critical genes at the breakpoint on chromosome 15 caused the Prader-Willi phenotype. FISH in a child with an Angelman phenotype showed no microdeletion, though Trisomy 15 was seen in 1 metaphase suggesting uniparental disomy due to trisomy rescue. A known polymorphism in the form of an additional tiny green signal on chromosome 14 was observed in 17 of 284 (6%) cases studied for Prader-Willi/Angelman syndrome. Another inherited polymorphism was seen in 5 cases, where one control signal was very small. Prenatal diagnosis was carried out with normal results, in 12 women with a previously affected child.