

Heterozygous Complete *NIPBL* Gene Deletion in Cornelia de Lange Syndrome: First Case Report from India

Shailesh Bajaj¹, Sheela Nampoothiri², Dhanya Yesodharan², Prakash Gambhir³ and Suvidya Ranade^{1*}

¹Department of Chemistry, Biochemistry Division, Savitribai Phule Pune University, Pune 411 007, Maharashtra, India

²Amrita Institute of Medical Sciences & Research Center, Cochin, Kerala, India

³Birthright Genetic Clinic, Erandwane, Pune, Maharashtra, India

KEYWORDS Cornelia de Lange Syndrome. MLPA. *NIPBL*. RT PCR

ABSTRACT Cornelia de Lange Syndrome-1 (CdLS; OMIM # 122470) is a multisystem, congenital, developmental disorder caused by heterozygous mutation in *NIPBL* gene on chromosome 5p13. CdLS is characterized by growth and developmental delay, facial dysmorphism, limb abnormality and other organ defects. The condition is mainly caused due to mutation in one of the cohesin ring forming genes. Among *NIPBL*, *SMC1A* and *SMC3*; *NIPBL* is mainly responsible for causing CdLS. To date molecular data for Indian CdLS patients is not available. Entire *NIPBL* gene has been screened in 12 children showing CdLS using MLPA in this study. The study reports entire gene deletion in one proband and partial gene deletion in the second proband. The observed deletion was in heterozygous condition in both the cases. The finding was validated by real time PCR.