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THE MECP2 DUPLICATION SYNDROME: WHAT'S NEW?

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In 2005, we showed that not only a loss of function of the *MECP2* gene but also a gain in *MECP2* dosage results in a severe neurodevelopmental phenotype in males. Patients with a duplication of chromosome region Xq28, that includes the *MECP2* gene, have severe mental retardation, facial and axial hypotonia, progressive spasticity, seizures and recurrent respiratory infections, which often leads to premature death. Since our initial description, more than 80 patients with a duplication of the *MECP2* gene have been described in the literature, broadening the phenotype. Adding 15 new and unpublished cases, enables us to give an update on what is currently known about the clinical phenotype, possible genotype-phenotype correlations and the mechanisms underlying this apparently frequent genomic aberration.