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Comparatively few reports exist describing the phenotype of Rubinstein-Taybi because of EP300 mutations. Clinical and genetic data were obtained from nine patients from the UK and Ireland with pathogenic EP300 mutations, identified either by targeted testing or by exome sequencing. All patients had mild or moderate intellectual impairment. Behavioural or social difficulties were noted in eight patients, including three with autistic spectrum disorders. Typical dysmorphic features of Rubinstein-Taybi were only variably present. Additional observations include maternal pre-eclampsia (2/9), syndactyly (3/9), feeding or swallowing issues (3/9), delayed bone age (2/9) and scoliosis (2/9). Six patients had truncating mutations in EP300, with pathogenic missense mutations identified in the remaining three. The findings support previous observations that microcephaly, maternal pre-eclampsia, mild growth restriction and a mild to moderate intellectual disability are key pointers to the diagnosis of EP300-related RTS. Variability in the presence of typical facial features of Rubinstein-Taybi further highlights clinical heterogeneity, particularly among patients identified by exome sequencing. Features that overlap with Floating-Harbor syndrome, including craniofacial dysmorphism and delayed osseous maturation, were observed in three patients. Previous reports have only described mutations predicted to cause haploinsufficiency of EP300, whereas this cohort includes the first described pathogenic missense mutations in EP300.
Rubinstein-Taybi syndrome type 2: report of nine new cases that extend the phenotypic and genotypic spectrum.

Hamilton. Clinical Dysmorphology (2016) ISSN: 0962-8827 Online ISSN: 1473-5717

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