De Novo COL7A1 mutation in a patient with trisomy 21: Coexistence of dystrophic epidermolysis bullosa and Down syndrome

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Background Down syndrome (DS) is the most common autosomal chromosomal disorder. Epidermolysis bullosa (EB) is a rare genodermatosis characterized by skin and mucous membrane fragility, with formation of blisters and erosions after minor trauma. Dystrophic EB (DEB) is inherited as an autosomal dominant (DDEB) or recessive (RDEB) trait. Both forms are caused by mutations in COL7A1, the gene coding for the type VII collagen. We report a patient affected by both conditions: DS and DDEB. Methods A patient with DS developed generalized blisters at the age of threemonths. Cytogenetic study was performed to confirm DS. Skin biopsies were examined with immunohistochemical and electron microscopy techniques to determine EB subtype. Genomic DNA was extracted from peripheral blood samples. COL7A1 mutations were screened by heteroduplex analysis using conformation-sensitive gel electrophoresis and sequencing. Results Karyotype analysis revealed trisomy 21. Histological study agreed with a DEB diag