pruriginosa、台湾人では通常の優性栄養障害型であり、VII 型コラーゲンの基本的な部分での塩基配列に差異はみとめられなかった。VII 型コラーゲンのそれ以外の部位や他の遺伝子も、栄養障害型表皮水疱症の症状に関与していることが推測された。

表皮水疱症において、プレクチンの変異では通常筋ジストロフィーを伴う単純型、アルファ6やベータ4インテグリンの変異で幽門閉鎖を伴う接合部型が発症する。当初、今回の症例では、幽門閉鎖のある接合部型を考えたが、電顕では表皮細胞レベルの水疱で、単純型を示唆するものであった。また、蛍光抗体法でプレクチンの発現が見られなかった。そこで、患者でのプレクチンの変異検索を行い、プレクチン遺伝子の変異を検出した。今回経験した2症例は、プレクチンの変異で起こる幽門閉鎖を伴う単純型で、表皮水疱症の新病型と考えた。

E 結論

今回行なった検索でも、表皮水疱症患者の遺伝子変異と臨床症状の関連が明確にならない部分も多く、さらに多くの症例の解析が必要と思われる。

F 健康危険情報

特になし。

G 研究発表

論文1) The G2028R glycine substitution mutation in *COL7A1* leads to marked inter-familiar clinical heterogeity in dominant dystrophic epidermolysis bullosa. Nakamura H, Sawamura D, Goto M, Sato-Matsumura KC, LaDuca J, Lee JYY, Masunaga T, Shimizu H. J Dermaol Sci, in press.

その他、投稿準備中。

H 知的財産の出願・登録状況

特になし。

III. 研究成果の刊行に関する一覧表

研究成果の刊行に関する一覧表（雑誌）

発表者氏名	論文タイトル名	発表誌名	巻号	ページ	出版年
Nakamura H, Sawamura D, Goto M, Sato-Matsumura KC, LaDuca J, Lee JYY, Masunaga T, Shimizu H	The G2028R glycine substitution mutation in *COL7A1* leads to marked inter-familiar clinical heterogeity in dominant dystrophic epidermolysis bullosa	**J Dermaol Sci**			in press
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以降は雑誌/図書等に掲載された論文となりますので、
「研究成果の刊行に関する一覧表」をご参照ください。