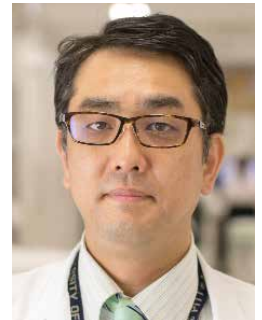


The Periodic diseases: JanewayとPAAND ～Generalistも遭遇する希少疾患～

The periodic diseases: Janeway and PAAND
～ rare diseases that generalists also encounter

飯塚病院総合診療科
清田 雅智



Masatomo Kiyota

Department of General Internal Medicine, Iizuka Hospital

対象者 医師, 若手医師 (卒後 16 年以内), 後期研修医 (卒後 3 年以上), 初期研修医 (卒後 1-2 年目), 学生

Target Doctor, Early Career Physicians, Senior Resident, Resident (1-2 years after graduation), Medical Student

Theodor Caldwell Janeway(1872-1917)は、感染性心内膜炎のJaneway's lesionの報告者Gamaliel Janeway(1841-1911)の息子で、Johns Hopkins Hospitalの内科教授である。1908年に彼はFamilial Mediterranean fever (FMF)の症例報告を行っていた。著者は、同様に腹痛と発熱を繰り返す45歳の男性を経験した。当時FMFを疑って久留米大学にMEFV遺伝子の依頼を行ったが、通常のexon 10には異常がなく、exon 2に異常のある希少疾患PAAND (pyrin-associated autoinflammation with neutrophilic dermatosis)と判明し、これは国内の最初の報告例であった。一般医でも、希少疾患への貢献ができることを示すことを発表したい。

Theodor Caldwell Janeway (1872-1917) is the son of Gamaliel Janeway (1841-1911), a reporter of Janeway's lesions in infective endocarditis, and a professor of internal medicine at Johns Hopkins Hospital. In 1908 he reported a case of Familial Mediterranean fever (FMF). I encountered a 45-year-old man who also had repeated abdominal pain and fever. At that time, I suspected FMF and requested the MEFV gene testing for Kurume University, but it turned out to be a rare disease PAAND (pyrin-associated autoinflammation with neutrophilic dermatosis) with abnormality in exon 2, without in exon 10. This is the first reported case in Japan. I would like to announce that even general practitioners can contribute to rare diseases.