

The 6th Joint Conference Ramathibodi - Osaka University

~ Cutting Edge Innovation on COVID-19 and Beyond ~

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Abstract Sheet

First Name/Last Name	Ken-ichi Hirano 平野 賢一
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Abstract 【English】

Triglyceride deposit cardiomyovasculopathy (TGCV) is a novel rare disease entity we found in Japanese patients with heart failure requiring cardiac transplantation (N Engl J Med. 2008). In TGCV, the defective intracellular lipolysis results in adult-onset heart failure and diffuse narrowing coronary artery disease resistant to standard remedies including cholesterol-lowering therapy and percutaneous coronary intervention. Recently, TGCV was encoded as a new orphan disease in the Orphanet (ORPHA code: 565612). The cumulative patient number so far diagnosed in Japan is 336. Fifty-eight of them have already died, indicating high risk profile. As similar to those with other orphan diseases, TGCV patients have been suffering from the unawareness, un-diagnosis, and delayed diagnosis. In this lecture, we present the diagnostic criteria 2020 of TGCV to facilitate the diagnosis inside and outside Japan and academia-initiated development of a drug for TGCV (coded as CNT-01). We look forward to global collaboration to save TGCV patients.

Abstract 【日本語】

中性脂肪蓄積心筋血管症 (TGCV) は、我々が心臓移植待機症例から見出した新規疾患単位である (N Engl J Med. 2008) (ORPHA code: 565612)。TGCVでは、血管平滑筋細胞、心筋細胞などにおける細胞内TG分解障害に起因して、患者は成人発症、治療抵抗性の心不全、びまん性冠動脈硬化症に悩む。最新の累積診断数は336例、内 58例が既に死亡している。本講演では、厚生労働省や日本医療開発研究機構の難病事業として行ってきたTGCV診断基準2020の策定や2020年に先駆け指定医薬品となったTGCV治療薬CNT-01 のアカデミア開発について述べる。