

厚生労働科学研究費補助金（難治性疾患政策研究事業）
分担研究報告書

性分化・性成熟異常を伴う内分泌症候群
(プラダーウイリ症候群・ヌーナン症候群を含む) の診療水準向上を目指す調査研究

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研究要旨

本研究の目的は、性分化・性成熟異常を伴う内分泌症候群であるプラダーウイリ症候群(PWS) (指定難病 193)、ヌーナン症候群(NS) (指定難病 195)、ターナー症候群(TS)、マッキューンオルブライト症候群(MAS)、バルデビードル症候群(BBS)、多嚢胞性卵巣症候群(PCOS) (全て小児慢性特定疾病) の医療水準向上のために診断基準、ガイドライン、疾患レジストリー構築を行った。

A. 研究目的

本研究の目的は、性分化・性成熟異常を伴う内分泌症候群の医療水準向上である。

B. 研究方法

プラダーウイリ症候群(PWS) (指定難病193)、ヌーナン症候群(NS) (指定難病195)、ターナー症候群(TS)、マッキューンオルブライト症候群(MAS)、バルデビードル症候群(BBS)、多嚢胞性卵巣症候群(PCOS) (全て小児慢性特定疾病) の診断基準、ガイドライン、疾患レジストリー構築。
(倫理面への配慮)
特になし

C. 研究結果

上記疾患の診断基準、ガイドライン、疾患レジストリー構築をおこなった。

D. 考察

研究分担者は内分泌内科医であり、成人診療の立場からトランジション、成人後のフォロー等を含めて議論に参加した。

E. 結論

当初の目標を達成した。

F. 健康危険情報

特になし

G. 研究発表

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previously treated patients with AGHD
The 12th Emirates Diabetes & Endocrine
Congress 2022 Virtual
(発表誌名巻号・頁・発行年等も記
入)

H. 知的財産権の出願・登録状況
(予定を含む。)

1. 特許取得
なし
2. 実用新案登録
なし
3. その他
特になし