資料９
新たな遺伝子異常に起因する先天性副腎低形成症の論文リスト

本研究班の代表・分担研究者をアンダーラインで示す

「SAMD9 異常に起因する先天性副腎低形成症」

SAMD9 mutations cause a novel multisystem disorder, MIRAGE syndrome, and are associated with loss of chromosome 7.
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Reversion SAMD9 Mutations Modifying Phenotypic Expression of MIRAGE Syndrome and Allowing Inheritance in a Usually de novo Disorder.
Roucher-Boulez F, Mallet D, Chatron N, Dijoud F, Gorduza DB, Bretones P, Morel Y. Front Endocrinol (Lausanne). 2019 Sep 11;10:625.

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Buonocore F, Achermann JC. Clin Endocrinol (Oxf). 2020 Jan;92(1):11-20.

A Rare Etiology of 46,XY Disorder of Sex Development and Adrenal Insufficiency: A Case of MIRAGE Syndrome Caused by Mutations in the SAMD9 Gene.
Mengen E, Küçükçongar Yavaş A, Uçaktürk SA. J Clin Res Pediatr Endocrinol. 2020 Jun 3;12(2):206-211.

「SGPL1 異常に起因する先天性副腎低形成症」

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