

形などの LDS と似た表現型を持った個体が認められた。

今後、この LDS モデルフィッシュを用いて、TGFβ シグナルと LDS の病態との関連を解明していく予定である。

2-4.

Different phenotypes in three patients with SCN2A variant

(大学院博士課程 4 年小児科・思春期科学分野)

○渡邊 由祐

(小児科・思春期科学分野)

森地振一郎、山中 岳、加納佳奈子

高松 朋子、春日 晃子、竹下 美佳

森下那月美、石田 悠、小穴 信吾

河島 尚志

(昭和大学医学部小児科学講座)

加藤 光広

SCN2A encodes voltage-gated sodium channel subunit alpha Nav1.2 expressed in the brain, especially in humans. Mutations of SCN2A can cause various neurodevelopmental disease including epilepsy, intellectual disability and developmental disorder.

(Case presentation) Case 1 (4-year-old male) : Apnea and asymmetric tonic posturing occurred since newborn period. Rhythmic sharp-and-spike waves in the left frontal and temporal lobes were noted on an electroencephalogram (EEG), for which treatment with PB and MDL was started at 4 days old. The SCN2A mutation c.4718T>C (p.L1573P) was confirmed by genetic testing. VPA and diazepam were ineffective, but lidocaine provided sufficient control. Eventually, the seizures disappeared without medication. He has moderate developmental delay. Case 2 (2-year-old male) : Respiratory disturbance developed after birth, with systemic tonic seizures occurring at 8 days old. An EEG revealed highvoltage slow waves. The SCN2A mutation c.4780T>C (p.Trp1594Arg) was confirmed by genetic testing. The symptoms were controlled with CBZ, CZP, ZNS and LID, although severe developmental delay persisted. Case 3 (8-month-old girl) : Systemic tonic seizures occurred at 1 day old and spasms

developed. An EEG revealed a suppression-burst pattern and the patient was diagnosed with Ohtahara syndrome at 3 months old. The SCN2A mutation c.4782G>C (p.Trp1594Cys) was confirmed by genetic testing. The symptoms were controlled with PB and mexiletine, although severe developmental delay persisted.

(Conclusion) These patients exhibited close mutation sites, although their neurological prognoses were different. As hypoxic ischemic encephalopathy and Ohtahara syndrome were noted in Cases 2 and 3, respectively, other neurological complications need to be explored with reference to previous reports.

2-5.

病原糸状菌 *Aspergillus fumigatus* の血清存在下での増殖に関する因子の同定

(微生物学分野)

○犬飼 達也、柴田 岳彦、宮崎 治子

大楠 清文、中村 茂樹

(国立感染症研究所：真菌部)

宮崎 義継

【背景・目的】 糸状真菌 *Aspergillus fumigatus* が主に引き起こす肺アスペルギルス症は、適切な治療薬・治療法を施してもなお高い致死率である。さらに、本菌の薬剤耐性菌の出現が世界各国から報告されている状況を踏まえれば、現状でも数の少ない既存抗真菌薬を用いた治療では、治療が難渋する可能性が予想される。この問題を解決する一つには、新規治療法、治療薬の開発が急務であり、真菌特異的な新たな標的や制御法を見出す必要がある。本菌は、他のアスペルギルス属とは異なり、血清存在下での増殖能が優れており、その表現型が、病原性や病態形成に寄与する可能性も示唆されているため、本菌の血清存在下での増殖に関する因子の探索を行った。

【方法】 血清存在下での増殖に影響を及ぼす菌側の因子を特定する目的で、分泌タンパク質をコードする遺伝子の破壊株を用い、血清存在有無の条件で培養後、増殖能をクリスタルバイオレット法により評価した。血清存在下で影響が認められた B11b 遺伝子破壊株と親株とを DNA マイクロアレイにより、

遺伝子発現の変化を比較し、血清存在下での増殖に関与するさらなる遺伝子の特定を行った。

【結果・考察】 親株において血清添加による発現量の増加が確認された遺伝子 543 個のうち、遺伝子破壊株において発現量の増加が観察されなかった 111 個の遺伝子について、遺伝子破壊株の表現型解析を行い、血清存在下での増殖に関与すると予想される遺伝子を 3 個特定した。特定した 3 遺伝子それぞれの破壊株を用いて、マウス感染実験を行った結果、全ての破壊株感染マウス群において、親株感染マウス群と比べて、顕著な生存率の上昇が確認された。以上より、特定した 3 つの遺伝子が血清存在下での菌糸生育を可能にし、感染宿主における増殖機構に関与することが示唆された。今後、これまでに特定した血清存在下の生育に必須な因子間の相互関係を明らかにし、血清存在下の増殖機構の解明を目指す。

2-6.

Treatment of intra-anal warts with imiquimod 5% cream : a single center prospective open study

(皮膚科学分野)

○入澤 亮吉、坪井 良治、齋藤万寿吉

原田 和俊

【Objectives】 Intra-anal warts are frequently recalcitrant to surgical removal, but imiquimod 5% cream is not formulated to use in medical care practice due to the risk of mucosal inflammation. In the present, single center, prospective, open study, we examined the safety and efficacy of application of imiquimod 5% cream for intra-anal warts.

【Methods】 Imiquimod 5% cream was applied to the entire inner surface of the anal canal with a cotton swab under anoscopy three times weekly for 16 weeks. If complete remission was not achieved, the treatment was continued until week 28. Electrocautery was applied once in a poorly responsive case.

【Results】 21 patients with intra-anal warts, of whom 16 were HIV-positive, were enrolled. Two patients withdrew before week 16, and nine more patients withdrew before week 28. The complete clearance rate was 36.8% (7/19) at week 16, and 70% (7/10) at week 28. Four patients achieved complete clearance at week

16 maintained clearance at week 28 without further treatment. Three out of 4 patients resistant to previous electrocautery achieved clearance with imiquimod 5% cream treatment. Adverse events occurred in 81% (17/21) of the patients mainly at the application site, but serious or previously unencountered adverse events were not observed.

【Conclusion】 Imiquimod 5% cream applied to intra-anal warts was nearly as efficacious and safe as when applied to external anogenital warts. Since treatment modalities for intra-anal warts are very limited, application of imiquimod 5% cream alone with careful and frequent observation or in combination with electrocautery is a useful option for refractory cases of intra-anal wart.

3-①-1.

Preliminary Findings on Control of Dispersion of Aerosols and Droplets During Bronchoscopy Using a newly developed oxygen Mask (Universal use of closed face oxygen masks for protect against developing COVID-19)

(社会人大学院博士課程 3 年呼吸器・甲状腺外科)

○田中 裕紀

(呼吸器・甲状腺外科)

濱中和嘉子、垣花 昌俊、河口 洋平

山田 祐揮、嶋田 善久、前原 幸夫

萩原 優、大平 達夫、池田 徳彦

Background: Considering the global spread of COVID-19 infection, routinely performed aerosol generating procedures such as bronchoscopy have been associated with infection of health-care workers. New strategy for the safe bronchoscopic examination should be created. This is the initial report of a study using computational fluid dynamic simulation to evaluate the newly modified oxygen mask during bronchoscopy.

Methods: The effectiveness of the modified oxygen mask was investigated by visualizing droplets and aerosols. And we quantified and compared droplets and aerosols with and without the newly mask. The airborne droplets counting was performed at two points, 60 cm and 30 cm from the mouth, assuming the distance