瘍を呈する症候群として認識される。一部で突然変異例がある。

Souza らは、19p13.3 の 1.1Mb 欠失によって発症した Peutz-Jeghers 症候群例を報告した。欠失領域には STK11 を含む 47 遺伝子が含まれ、知的障害、特異顔貌、先天性心疾患を合併した。Scollon らも、19p13.3 欠失による Peutz-Jeghers 症候群に加えて発達遅滞、粘膜下口蓋裂、てんかん発作を伴う小児例を報告した。

Peutz-Jeghers 症候群を伴う 19p13.3 欠失は、まだ症候群として認知されるには至っていないが、Peutz-Jeghers 症候群に加えて、精神運動発達遅滞や特異顔貌、口蓋裂などの共通した所見を認める。欠失が突然変異であるので、当然 Peutz-Jeghers 症候群は孤発性である。家族例のない Peutz-Jeghers 症候群で発達遅滞その他の合併症を認めた場合、19p13.3 欠失を疑ってマイクロアレイ染色体検査を行う必要がある。変異解析では異常がみつからない。

症例3で合併した自閉症の責任遺伝子もこの欠失範囲内に存在すると考えられたが、特定に至っておらず、今後の症例の蓄積が必要と考えられた。

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(3)症例4について

RASopathies は、Ras/MAPK 系の異常による先天異常症候群であり、Noonan 症候群、CFC 症候群、Costello 症候群などが該当する。19p13.3 には、CFC 症候群の責任遺伝子のひとつである *MEK2*(MAPK/ERK KINASE 2)が存在する。CFC 症候群における MEK2 変異の種類は基本的にはミスセンス変異である。最近 Nowaczyk らは、*MEK2* を含む欠失により、CFC 症候群類似の所見を呈する例を報告した。顔貌は、高い前額、厚い鼻先、未発達の頬骨、長い顔面正中部、角張った下顎、顔面非対照などの特徴を認めた。発達遅滞、筋緊張低下、体重増加不良、心疾患、閉塞性睡眠時無呼吸、胃食道逆流などの合併症を認めた。従来、RASopathies は、Ras/MAPK 系の機能獲得変異が原因といわれていたが、*MEK2* 欠失も類似の所見を呈することが明らかになった。

症例4では *MEK2* を含む微細欠失を認めた。臨床的にはCFC症候群とは異なるが、*MEK2* を含む複数の遺伝子の欠失が知的障害の原因と考えられた。

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E. 結論

19 番染色体の微細欠失症候群として、19p13.3 欠失症候群は Sotos 症候群2型の責任遺伝子である *NFIX* を含む新規症候群として確立されつつある。

NFIX よりもややセントロメアよりに位置する *STK11* 遺伝子を含む欠失は Peutz-Jeghers 症候群を合併する。Peutz-Jeghers 症候群に加えて、特異顔貌、精神運動発達遅滞を合併する。Peutz-Jeghers 症候群の孤発例の中に本症候群が含まれる可能性があり、臨床的に重要と考えられた。

CFC 症候群は従来、RAS-MAPK 系に関わる特定の遺伝子の機能獲得変異が原因とされたが、MEK2 の欠失も知的障害を呈することが示された。

19 番染色体の微細欠失の症例数は少ないが、認識可能な症候群として確かに存在すると考えられた。こうした疾患の診断にはマイクロアレイ染色体検査が必須である。

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2. 著書

なし

3. 学会発表

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H. 知的所有権の取得状況

1. 特許取得

なし

2. 実用新案登録

なし

3. その他

Ⅲ. 研究成果の刊行に関する一覧表

研究成果の刊行に関する一覧表

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