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Università Roma Sapienza

Polo Pontino – ICOT Latina

Nuove metodologie di analisi genetica: Successi e problematiche - 2

Pannello geni HSP – Fondazione Stella Maris – Pisa – Dott. Santorelli

117 geni !

Data accettazione: 19/12/2013

Data di inizio dell'esecuzione del test: 06/06/2018

Provenienza: Prof. C. Casali, Neurologia, Università Sapienza, Polo Pontino, Latina

Indicazione clinica all'indagine: Paraparesi spastica ereditaria

Materiale esaminato: DNA

Metodica utilizzata: Target re-sequencing con piattaforma MiSeq Illumina. Le sonde utilizzate per l'esperimento sono state disegnate secondo la tecnologia SureSelect, con ibridazione bridge PCR

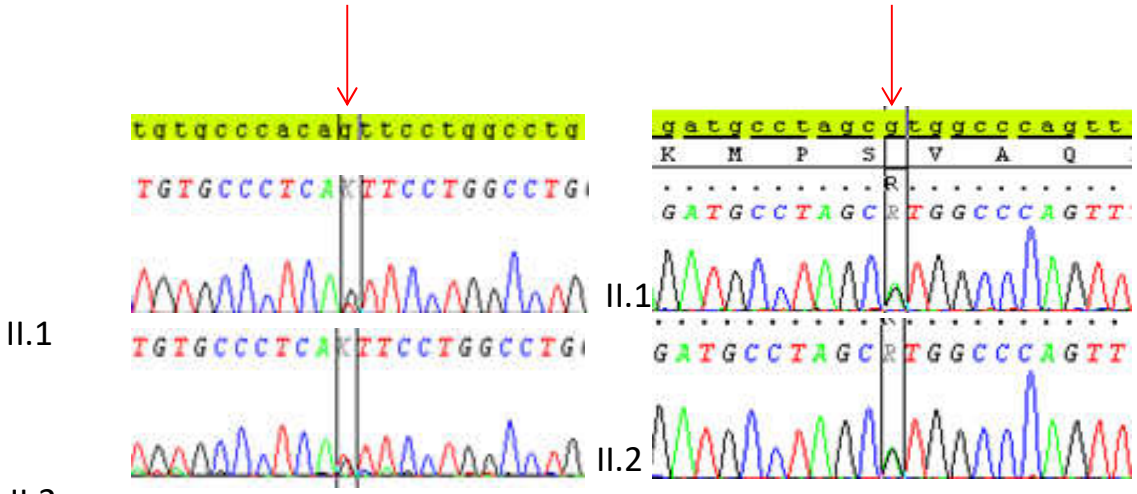
Sequenziamento degli esoni codificanti dei seguenti geni: *ABCD1, ACBD5, ADAR , AFG3L2, ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATAD3A, ATL1, ATL3, ATP13A2, ATP2B4, B4GALNT1, BICD2, BSCL2, C12ORF65, C19ORF12, C9ORF72, CAPN1, CCT5, COASY, CPT1C, CYP27A1, CYP2U1, CYP7B1, DARS2, DDHD1, DDHD2, DSTYK, DYNC1H1, ENTPD1, EPT1, ERLIN1, ERLIN2, EXOSC3, EXOSC8, FA2H, FARS2, FBXO7, FLRT1, GAD1, GBA2, GCH1, GDAP1, GDAP2, GJA1, GJC2, GLB1, HSPD1, IBA57, IFIH1, KCNA2, KIAA0196, KIF1A, KIF1C, KIF5A, KY, L1CAM, LYST, MAG, MARS, MARS2, MFN2, MTHFR, MTPAP, NIPA1, NPC1, NT5C2, OPA1, OPA3, PANK2, PEX16, PGAP1, PLA2G6, POLR3A, RAB3GAP2, REEP1, REEP2, RNASEH2B, RTN2, SACS, SAMHD1, SERAC1, SETX, SLC16A2, SLC33A1, SOX10, SPAST, SPG11, SPG20, SPG21, SPG7, TECPR2, TFG, TRMT5, TRPV4, TTC19, TUBB4A, UBQLN2, UCHL1, USP8, VAMP1, VCP, VPS37A, VRK1, WDR48, ZFR, ZFYVE26, ZFYVE27*

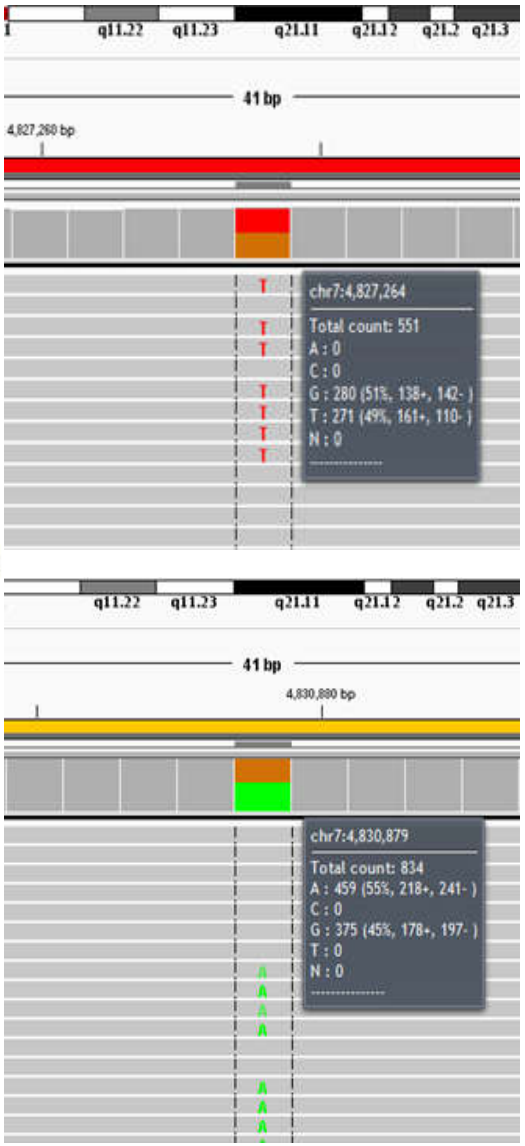
Risultati: L'analisi eseguita ha identificato le varianti di significato patogenetico incerto riportate in tabella.

Commenti: I dati ottenuti vanno interpretati nel contesto del quadro clinico e nell'ambito della consulenza genetica allargata.

Fam St- *AP5Z1/SPG48*

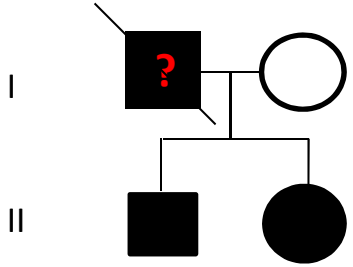
mut in *AP5Z1* : c.1312-1G>T + c.2287G>A (p.V763M)





▼ Inheritance

The transmission pattern of SPG48 in the family reported by Slabicki et al. (2010) was consistent with autosomal recessive inheritance.



NGS data from patient II.2

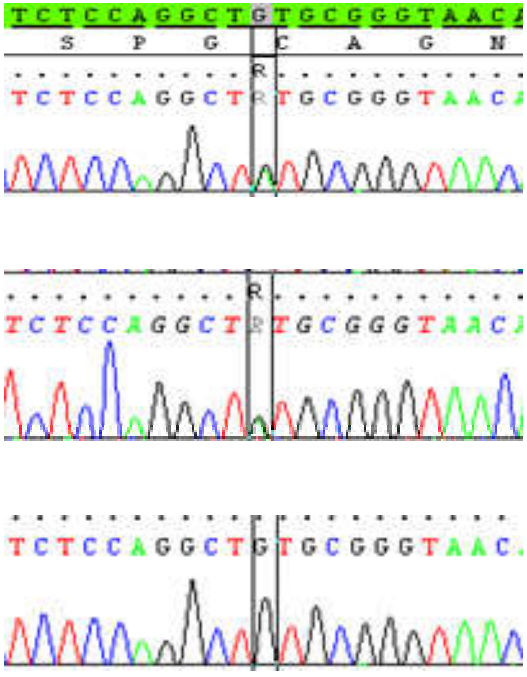
Fam Z – *ERLIN2/SPG18*

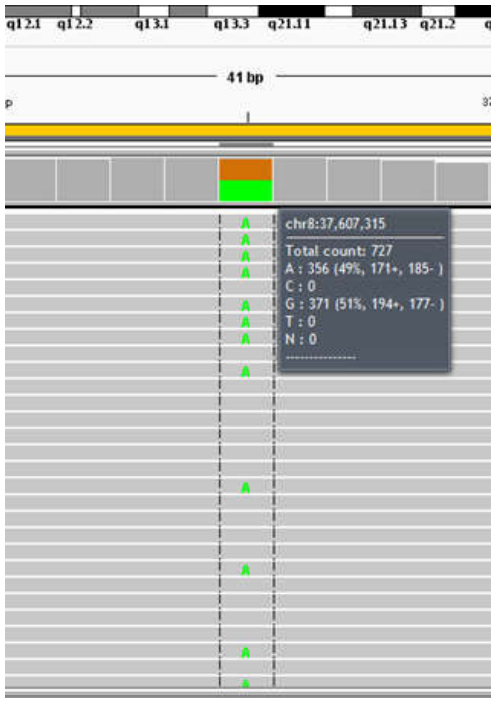
mut c.502G>A (p.V168M) – *ERLIN2*

I.2

II.1

II.2

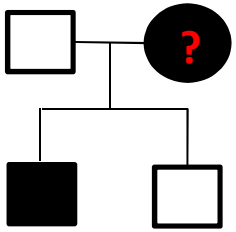




NGS data from patient II.1

I

II



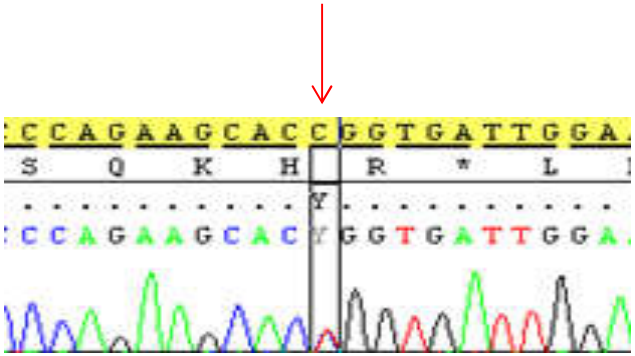
A novel heterozygous variant in *ERLIN2* causes autosomal dominant pure hereditary spastic paraplegia

S. L. Rydning[a,b,*], A. Dudesek[c,d,*], F. Rimmele[c,d], C. Funke[e], S. Krüger[e], S. Biskup[e,f], M. D. Vigeland[g], H. S. Hjorthaug[g], Y. Sejersted[g], C. Tallaksen[a,b], K. K. Selmer[a,g] and C. Kamm[c]

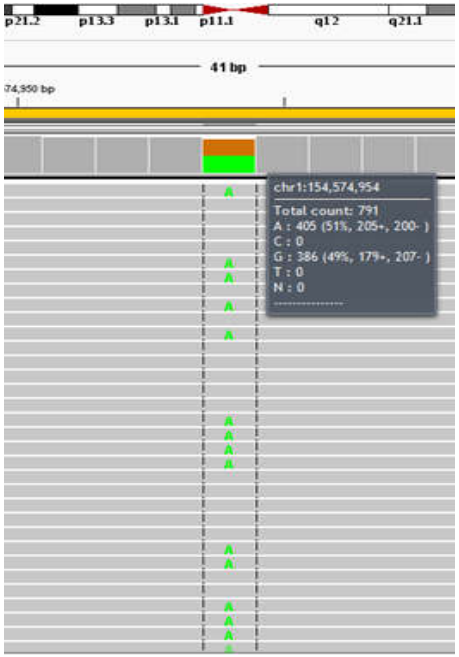
[a]*Institute of Clinical Medicine, University of Oslo, Oslo;* [b]*Department of Neurology, Oslo University Hospital, Oslo, Norway;* [c]*Department of Neurology, University of Rostock, Rostock;* [d]*German Center for Neurodegenerative Diseases (DZNE), Rostock, Germany;* [e]*CeGaT GmbH, Center for Genomics and Transcriptomics, Tübingen;* [f]*Hertie-Institute for Clinical Brain Research and German Center for Neurodegenerative Diseases (DZNE), University of Tübingen, Tübingen, Germany; and* [g]*Department of Medical Genetics, Oslo University Hospital, Oslo, Norway*

Fam L – *ADAR*

mut. c.164C>T (p.P55L) – *ADAR*



No relatives for segregation studies.



NGS data

225750

AICARDI-GOUTIERES SYNDROME 1; AGS1

Alternative titles; symbols

AGS
ENCEPHALOPATHY, FAMILIAL INFANTILE, WITH INTRACRANIAL CALCIFICATION AND CHRONIC CEREBROSPINAL FLUID LYMPHOCYTOSIS
CREE ENCEPHALITIS
PSEUDOTOXOPLASMOSIS SYNDROME

Mutations in *ADAR1*, *IFIH1*, and *RNASEH2B* Presenting As Spastic Paraplegia

Yanick J. Crow[1,2,3] Maha S. Zaki[4] Mohamed S. Abdel-Hamid[5] Ghada Abdel-Salam[4] Odile Boespflug-Tanguy[6,7] Nuno J. V. Cordeiro[8] Joseph G. Gleeson[9] Nirmala Rani Gowrinathan[10] Vincent Laugel[11] Florence Renaldo[12,13,14] Diana Rodriguez[15] John H. Livingston[16] Gillian I. Rice[3]

L’ansia da diagnosi

- Ansia del medico e del paziente
- Cosa fare per pazienti con e senza diagnosi:
  - Gestione clinica globale
  - Informazione e formazione
  - Riabilitazione

Presa in carico

- Attività fondamentale
- Diagnosi clinica e genetica
- Riabilitazione come processo globale
  - Fisioterapia
  - Riabilitazioni speciali (logopedia, deglutizione, respiratoria)
  - Ortesi e ausili
  - Terapia occupazionale
  - Continuità assistenziale
  - Aspetti sociali e psicologici