

## Contents

### Epigenetics

- 793** When chromatin organisation floats astray: the *Srcap* gene and Floating-Harbor syndrome  
**[MCQs]** *G Messina, M T Atterato, P Dimiri*

### Commentary

- 798** Anxiety delivered direct-to-consumer: are we asking the right questions about the impacts of DTC genetic testing?  
*S Oliveri, H C Howard, C Renzi, M G Hansson, G Pravettoni*

### Cancer genetics

- 800** *PALB2*, *CHEK2* and *ATM* rare variants and cancer risk: data from COGS  
**[OPEN ACCESS]** *M C Southey, D E Goldgar, R Winqvist, K Pylkäs, F Couch, M Tischkowitz, W D Foulkes, J Dennis, K Michailidou, E J van Rensburg, T Heikkinen, H Nevanlinna, J L Hopper, T Dörk, K B M Claes, J Reis-Filho, Z L Teo, P Radice, I Caucci, P Peterlongo, H Tsimiklis, F A Odeh, J G Dowty, M K Schmidt, A Broeks, F B Hogervorst, S Verhoef, J Carpenter, C Clarke, R J Scott, P A Fasching, L Haeberle, A B Ekici, M W Beckmann, J Peto, I dos-Santos-Silva, O Fletcher, N Johnson, M K Bolla, E J Sawyer, I Tomlinson, M J Kerin, N Miller, F Marme, B Burwinkel, R Yang, P Guénel, T Truong, F Menegaux, M Sanchez, S Bojesen, S F Nielsen, H Flyger, J Benitez, M Pilar Zamora, J I A Perez, P Menéndez, H Anton-Culver, S Neuhausen, A Ziogas, C A Clarke, H Brenner, V Arndt, C Stegmaier, H Brauch, T Brüning, Y-D Ko, T A Muranen, K Aittomäki, C Blomqvist, N V Bogdanova, N N Antonenkova, A Lindblom, S Margolin, A Mannermaa, V Kataja, V-M Kosma, J M Hartikainen, A B Spurdle, leConFab Investigators, Australian Ovarian Cancer Study Group, E Wauters, D Smeets, B Beuselinck, G Floris, J Chang-Claude, A Rudolph, P Seibold, D Flesch-Janys, J E Olson, C Vachon, V S Pankratz, C McLean, C A Haiman, B E Henderson, F Schumacher, L Le Marchand, V Kristensen, G G Alnæs, W Zheng, D J Hunter, S Lindstrom, S E Hankinson, P Kraft, I Andrusis, J A Knight, G Glendon, A M Mulligan, A Jukkola-Vuorinen, M Grip, S Kauppila, P Devilee, R A E M Tollenaar, C Seynaeve, A Hollestelle, M Garcia-Closas, J Figueroa, S J Chanock, J Lissowska, K Czene, H Darabi, M Eriksson, D M Eccles, S Rafiq, W J Tapper, S M Gerty, M J Hooning, J W M Martens, J Margriet Collé, M Tilanus-Linthorst, P Hall, J Li, J S Brand, K Humphreys, A Cox, M W R Reed, C Luccarini, C Baynes, A M Dunning, U Hamann, D Torres, H U Ulmer, T Rüdiger, A Jakubowska, J Lubinski, K Jaworska, K Durda, S Slager, A E Toland, C B Ambrosone, D Yannoukakos, A Swerdlow, A Ashworth, N Orr, M Jones, A González-Neira, G Pita, M Rosario Alonso, N Álvarez, D Herrero, D C Tessier, D Vincent, F Bacot, J Simard, M Dumont, P Soucy, R Lees, K Muir, F Wiklund, H Gronberg, J Schleutker, B G Nordestgaard, M Weischer, R C Travis, D Neal, J L Donovan, F C Hamdy, K-T Khaw, J L Stanford, W J Blot, S Thibodeau, D J Schaid, J L Kelley, C Maier, A S Kibel, C Cybulski, L Cannon-Albright, K Butterbach, J Park, R Kaneva, J Batra, M R Teixeira, Z Kote-Jarai, A A Olama, S Benlloch, S P Renner, A Hartmann, A Hein, M Ruebner, D Lambrechts, E Van Nieuwenhuysen, I Vergote, S Lambrechts, J A Doherty, M A Rossing, S Nickels, U Eilber, S Wang-Gohrke, K Odunsi, L E Sucheston-Campbell, G Friel, G Lurie, J L Killeen, L R Wilkens, M T Goodman, I Runnebaum, P A Hillemanns, L M Peltari, R Butzow, F Modugno, R P Edwards, R B Ness, K B Moysich, A du Bois, F Heitz, P Harter, S Kommoss, B Y Karlan, C Walsh, J Lester, A Jensen, S K Kjaer, E Høgdall, B Peissel, B Bonanni, L Bernard, E L Goode, B L Fridley, R A Vierkant, J M Cunningham, M C Larson, Z C Fogarty, K R Kalli, D Liang, K H Lu, M A T Hildebrandt, X Wu, D A Levine, F Dao, M Bisogna, A Berchuck, E S Iversen, J R Marks, L Akushevich, D W Cramer, J Schildkraut, K L Terry, E M Poole, M Stampfer, S S Tworoger, E V Bandera, I Orlow, S H Olson, L Bjorge, H B Salvesen, A M van Altena, K K H Aben, L A Kiemeny, L F A G Massuger, T Pejovic, Y Bean, A Brooks-Wilson, L E Kelemen, L S Cooke, N D Le, B Górski, J Gronwald, J Menkiszak, C K Høgdall, L Lundvall, L Nedergaard, S A Engelholm, E Dicks, J Tyrer, I Campbell, I McNeish, J Paul, N Siddiqui, R Glasspool, A S Whittemore, J H Rothstein, V McGuire, W Sieh, H Cai, X-O Shu, R T Teten, R Sutphen, J R McLaughlin, S A Narod, C M Phelan, A N Monteiro, D Fenstermacher, H-Y Lin, J B Permuth, T A Sellers, Y Ann Chen, Y-Y Tsai, Z Chen, A Gentry-Maharaj, S A Gayther, S J Ramus, U Menon, A H Wu, C L Pearce, D Van Den Berg, M C Pike, A Dansonka-Mieszkowska, J Plisiecka-Halasa, J Moes-Sosnowska, J Kupryjanczyk, P D P Pharoah, H Song, I Winship, G Chenevix-Trench, G G Giles, S V Tavtigian, D F Easton, R L Milne*

### Copy-number variation

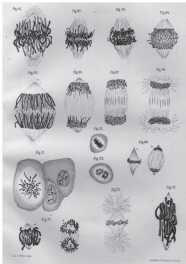
- 812** Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder  
*S J Mosca, L M Langevin, D Dewey, A M Innes, A C Lionel, C C Marshall, S W Scherer, J S Parboosingh, F P Bernier*

### Genotype-phenotype correlations

- 820** Eight further individuals with intellectual disability and epilepsy carrying bi-allelic *CNTNAP2* aberrations allow delineation of the mutational and phenotypic spectrum  
*M Smogavec, A Cleall, J Hoyer, D Lederer, M-C Nassogne, E E Palmer, M Deprez, V Benoit, I Maystadt, C Noakes, A Leal, M Shaw, J Gecz, L Raymond, A Reis, D Shears, K Brockmann, C Zweier*

December 2016 Volume 53 Issue 12

**JMG**  
Journal of Medical Genetics



[jmg.bmj.com](http://jmg.bmj.com)



**BMJ**

Cover credit: The classical 1882 illustration of mitosis by Walter Flemming, that gave chromatin its name (coloured substance). From Messina *et al.*, pg. 793.



Adopted as the official Journal of the Canadian College of Medical Geneticists

Receive regular table of contents by email. Register using this QR code.



This article has been chosen by the Editor to be of special interest or importance and is freely available online.



This article has been made freely available online under the BMJ Journals Open Access scheme. See <http://jmg.bmj.com/site/about/guidelines.xhtml#open>



This journal is a member of and subscribes to the principles of the Committee on Publication Ethics  
<http://publicationethics.org/>



When you have finished with this please recycle it

**[MCQs]** The online version of this article contains multiple choice questions hosted on BMJ Learning.

## Complex traits

- 828** Cystathionine  $\beta$ -synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations  
*L Goffinet, A Oussalah, R-M Guéant-Rodriguez, C Chery, M Basha, P H Avogbe, T Josse, E Jeannesson, P Rouyer, J Flayac, P Gérard, A Le Touze, B Bonin-Goga, D Goga, E Simon, F Feillet, M Viikula, J-L Guéant*

## Genome-wide studies

- 835** Meta-analysis of genome-wide association studies of HDL cholesterol response to statins  
*I Postmus, H R Warren, S Trompet, B J Arsenault, C L Avery, J C Bis, D I Chasman, C E de Keyser, H A Deshmukh, D S Evans, Q Feng, X Li, R A J Smit, A V Smith, F Sun, K D Taylor, A M Arnold, M R Barnes, B J Barratt, J Betteridge, S Matthijs Boekholdt, E Boerwinkle, B M Buckley, Y-D Ida Chen, A J M de Craen, S R Cummings, J C Denny, M P Dubé, P N Durrington, G Eiriksdottir, I Ford, X Guo, T B Harris, S R Heckbert, A Hofman, G K Hovingh, J J P Kastelein, L J Launer, C-T Liu, Y Liu, T Lumley, P M McKeigue, P B Munroe, A Neil, D A Nickerson, F Nyberg, E O'Brien, C J O'Donnell, W Post, N Poulter, R S Vasani, K Rice, S S Rich, F Rivadeneira, N Sattar, P Sever, S Shaw-Hawkins, D C Shields, P E Slagboom, N L Smith, J D Smith, N Sotoodehnia, A Stanton, D J Stott, B H Stricker, T Stürmer, A G Uitterlinden, W-Q Wei, R G J Westendorp, E A Whitsel, K L Wiggins, R A Wilke, C M Ballantyne, H M Colhoun, L A Cupples, O H Franco, V Gudnason, G Hüman, C N A Palmer, B M Psaty, P M Ridker, J M Stafford, C M Stein, J-C Tardif, M J Caulfield, J Wouter Jukema, J I Rotter, R M Krauss*

## Mitochondrial genetics

- 846** *COA7* (*C1orf163/RESA1*) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency  
*A M Lyons, A Ardisson, A Reyes, A J Robinson, I Moroni, D Ghezzi, E Fernandez-Vizarra, M Zeviani*



## New disease loci

- 850** De novo mutations of *KIAA2022* in females cause intellectual disability and intractable epilepsy  
*I M de Lange, K L Helbig, S Weckhuysen, R S Møller, M Velinov, N Dolzhanskaya, E Marsh, I Helbig, O Devinsky, S Tang, H C Mefford, C T Myers, W van Paesschen, P Striano, K van Gassen, M van Kempen, C G F de Kovel, J Piard, B A Minassian, M M Nezarati, A Pessoa, A Jacquette, B Maher, S Balestrini, S Sisodiya, M T A Warde, A De St Martin, J Chelly, EuroEPINOMICS-RES MAE working group, R van't Slot, L Van Maldergem, E H Brilstra, B P C Koeleman*



## Phenotypes

- 859** Novel asymptomatic CNS findings in patients with *ACVR1/ALK2* mutations causing fibrodysplasia ossificans progressiva  
*M Severino, M Bertamino, D Tortora, G Morana, S Uccella, R Bocciardi, R Ravazzolo, A Rossi, M Di Rocco*

## Correction

- 845** Erratum: The kinetochore protein, *CENPF*, is mutated in human ciliopathy and microcephaly phenotypes