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*Haematologica* 0 **102'** 0 **4P**

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*Eur J Med Genet* 0 **60'**

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E dentific tion of function y signific nt tri e ic genotype in t e yrosin se gene YR  
c using ypo orp ic ocu ocut neous inis OCA B *Nature Scientific Reports* 0  
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P ce NP **Maggouta F, Twigden M** Borg Mo ecu r cytogenetic c r cteris tion of no e *de*  
*novo* ring c ro oso e in o ing ter in p de etion nd ter in q dup ic tion in t e different  
r soft es e c ro oso e *Mol Cytogenet* 0 **0'**

R e G o Y **Ward D, Mattocks CJ** Eren E i i s AP A no e ger ine g in of function  
ri nt in P CD *Clin Immunology* 0 *Letter to the Editor* **181'**

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C et nR u rA n erited q icrode etions in o ing t e MBD ocus *Mol Genet &  
Genomic Medicine* 0 **5** 0

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Egger nn H r ison MD e p e **Mackay DJG** Netc ine Di gnosis nd n ge ent of  
Si er Russe syndro a first intern tion consensus st te ent *Nat Reviews Endocrinology* 0  
**13** 0 **40**

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er A B rtenstein M y s P **Cross NCP** M co ti L C zzo M He stro Lind erg E  
Og S S it C Pe g tti A Bou t ood e AF <sup>S405</sup> ut tion induces ine ge specific  
sp icing ter tions in ye odysp stic syndro es *J Clin Investigation* 0 **127** 0

Zoi nd **Cross NCP** Geno ics of ye opro ifer ti e neop s s *J Clin Oncology* 0 **35** **40**