Projectes finançats els darrers 6 anys

Títol del projecte: “Mecanismos causantes del síndrome de DiGeorge/Velo-Cardio-Facial y de las cardiopatías congénitas asociadas”.
Entitat financiadora: Instituto Salud Carlos III; Fondo Investigaciones Sanitarias (ref: 05-1585).
Entitats participants: 2
Durada, des de: 2005 fins: 2008
Quantitat de la subvenció: 54.740 €
Investigador responsable: Dr. Alexander Damián Heine Suñer
Nombre d’nvestigadors participants: 7

Títol del projecte: Intensificación investigadora.
Entitat financiadora: Instituto Salud Carlos III (ref: INT07/291).
Durada, 2008 Quantitat de la subvenció: 30000 €
Investigador beneficiari: Dr. Alexander Damián Heine Suñer

Títol del projecte: “Aproximación a la correlación genotipo-fenotipo y determinación de bases moleculares de las cardiopatías congénitas y del síndrome del22q11.2”.
Entitat financiadora: Instituto Salud Carlos III (ref: PI081363).
Durada, 2009-2011 Quantitat de la subvenció: 172425 €
Investigador responsable: Dr. Alexander Damián Heine Suñer
Nombre d' investigadors participants: 6

Títol del projecte: Intensificación investigadora.
Entitat financiadora: Instituto Salud Carlos III (ref: INT07/291).
Durada, 2009 Quantitat de la subvenció: 30000 €
Investigador beneficiari: Dr. Alexander Damián Heine Suñer

Títol del projecte: Análisis molecular de los puntos de rotura presentes en reorganizaciones cromosómicas
Entitat financiadora: Instituto Salud Carlos III (ref: CP07/00258).
Durada, 2008-2010 Quantitat de la subvenció: 40600€€
Investigador beneficiari: Dra. Maria Oliver Bonet

Títol del projecte: Influencia de los polimorfismos genéticos en la recombinación meiótica: relación con los síndromes genómicos y las reorganizaciones estructurales

Entitat financiadora: Instituto Salud Carlos III (ref: PI08/1185)

Durada, 2009-2011 Quantitat de la subvenció: 127.350€.

Investigador responsable: Dra. Maria Oliver Bonet

Nombre d' investigadors participants: 7

4) Publicacions

A continuació es detallen les publicacions dels darrers 6 anys dels membres del grup de Genòmica de la Salud:

1. Herman SB, Guo T, McGinn DM, Blonska A, Shanske AL, Bassett AS, Chow EW, Bowser M, Sheridan M, Beemer F, Devriendt K, Swillen A, Breckpot J, Digilio MC, Marino B, Dallapiccola B, Carpenter C, Zheng X, Johnson J, Chung J, Higgins AM, Philip N, Simon T, Coleman K, **Heine-Suner D**, Rosell J, Kates W, Devoto M, Zackai E, Wang T, Shprintzen R, Emanuel BS, Morrow BE; and the International Chromosome 22q11.2 Consortium.
   Overt cleft palate phenotype and TBX1 genotype correlations in velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients.. Am J Med Genet A. 2012 Nov;158A(11):2781-7.
2. García-Peiró A, **Oliver-Bonet M**, Navarro J, Abad C, Amengual MJ, López-Fernández C, Gosálvez J, Benet J.
   Differential clustering of sperm subpopulations in infertile males with clinical varicocele and carriers of rearranged genomes. J Androl. 2012 May-Jun;33(3):361-7.
3. Barca-Tierno V, Aza-Carmona M, Barroso E, **Heine-Suner D**, Azmanov D, Rosell J, Ezquieta B, Montané LS, Vendrell T, Cruz J, Santos F, Rodríguez JI, Pozo J, Argente J, Kalaydjieva L, Gracía R, Campos-Barros A, Benito-Sanz S, Heath KE.
   Identification of a Gypsy SHOX mutation (p.A170P) in Léri-Weill dyschondrosteosis and Langer mesomelic dysplasia. Eur J Hum Genet. 2011 Dec;19(12):1218-25.
4. García-Peiró A, **Oliver-Bonet M**, Navarro J, Abad C, Guitart M, Amengual MJ, Gosálvez J, Benet J.
   Dynamics of sperm DNA fragmentation in patients carrying structurally rearranged chromosomes. Int J Androl. 2011 Dec;34(6 Pt 2):e546-53.
5. Guo T, McDonald-McGinn D, Blonska A, Shanske A, Bassett AS, Chow E, Bowser M, Sheridan M, Beemer F, Devriendt K, Swillen A, Breckpot J, Digilio MC, Marino B, Dallapiccola B, Carpenter C, Zheng X, Johnson J, Chung J, Higgins AM, Philip N, Simon TJ, Coleman K, **Heine-Suner D**, Rosell J, Kates W, Devoto M, Goldmuntz E, Zackai E,

Wang T, Shprintzen R, Emanuel B, Morrow B; International Chromosome 22q11.2 Consortium.

Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. Hum Mutat. 2011 Nov;32(11):1278-89.

6. Rius M, Obradors A, Daina G, Ramos L, Pujol A, Martínez-Passarell O, Marquès L, **Oliver-Bonet M**, Benet J, Navarro J.

   Detection of unbalanced chromosome segregations in preimplantation genetic diagnosis of translocations by short comparative genomic hibridization. Fertil Steril. 2011 Jul;96(1):134-42.

7. García-Peiró A, **Oliver-Bonet M**, Navarro J, Abad C, Guitart M, Amengual MJ, Benet J.

   Sperm DNA integrity and meiotic behavior assessment in an infertile male carrier of a 9qh+++ polymorphism. J Biomed Biotechnol. 2011;2011:730847.

8. García-Pavía P, Avellana P, Bornstein B, **Heine-Suñer D**, Cobo-Marcos M, Gómez-Bueno M, Segovia J, Alonso-Pulpón LA.

   Familial approach in hereditary transthyretin cardiac amyloidosis. Rev Esp Cardiol. 2011 Jun;64(6):523-6.

9. Benito-Sanz S, Barroso E, **Heine-Suñer D**, Hisado-Oliva A, Romanelli V, **Rosell J**, Aragones A, Caimari M, Argente J, Ross JL, Zinn AR, Gracia R, Lapunzina P, Campos-Barros A, Heath KE.

   Clinical and molecular evaluation of SHOX/PAR1 duplications in Leri-Weill dyschondrosteosis (LWD) and idiopathic short stature (ISS). J Clin Endocrinol Metab. 2011 Feb;96(2):E404-12.

10. García-Peiró A, Martínez-Heredia J, **Oliver-Bonet M**, Abad C, Amengual MJ, Navarro J, Jones C, Coward K, Gosálvez J, Benet J.

    Protamine 1 to protamine 2 ratio correlates with dynamic aspects of DNA fragmentation in human sperm. Fertil Steril. 2011 Jan;95(1):105-9. Epub 2010 Jul 29.

11. Ruiz Plazas X, **Burgués Gasión JP**, Ozonas Moragues M, Pizá Reus P.

    Utility of inhibin B in the management of male infertility. Actas Urol Esp. 2010 Oct;34(9):781-7

12. Rius M, Obradors A, Daina G, Cuzzi J, Marquès L, Calderón G, Velilla E, Martínez-Passarell O, **Oliver-Bonet M**, Benet J, Navarro J.

    Reliability of short comparative genomic hybridization in fibroblasts and blastomeres for a comprehensive aneuploidy screening: first clinical application. Hum Reprod. 2010 Jul;25(7):1824-35.

13. Fernández L, Nevado J, Santos F, **Heine-Suñer D**, Martinez-Glez V, García-Miñaur S, Palomo R, Delicado A, Pajares IL, Palomares M, García-Guereta L, Valverde E, Hawkins F, Lapunzina P.

    A deletion and a duplication in distal 22q11.2 deletion syndrome region. Clinical implications and review. BMC Med Genet. 2009 Jun 2;10:48. Review.

14. López-Alvarez MR, Martínez-Sánchez MV, Salgado-Cecilia MG, Campillo JA, **Heine-Suñer D**, Villar-Permuy F, Fuster JL, Bas A, Gil-Herrera J, Muro M, García-Alonso AM, Alvarez-López MR, Minguela A.

    Association of monoclonal expansion of Epstein-Barr virus-negative CD158a+ NK cells secreting large amounts of gamma interferon with hemophagocytic lymphohistiocytosis. Clin Vaccine Immunol. 2009 Jan;16(1):142-5. Epub 2008 Nov 19.

15. Obradors A, Fernández E, Rius M, **Oliver-Bonet M**, Martínez-Fresno M, Benet J, Navarro

    Outcome of twin babies free of Von Hippel-Lindau disease after a double-factor preimplantation genetic diagnosis: monogenetic mutation analysis and comprehensive aneuploidy screening. J.Fertil Steril. 2009 Mar;91(3):933.e1-7.

16. Perrin A, Caer E, **Oliver-Bonet M**, Navarro J, Benet J, Amice V, De Braekeleer M, Morel F

    DNA fragmentation and meiotic segregation in sperm of carriers of a chromosomal structural abnormality. Fertil Steril. 2008 Aug 13. [Epub ahead of print]

17. Oliver-Bonet M.

    FISH on Sperm, Spermatocytes and Oocytes. In Fluorescence In Situ Hybridization (FISH) - Application Guide, Thomas Liehr (Ed.) Springer Protocols (en premsa) ISBN: 978-3-540-70580-2

18. Sun F, Mikhaail-Philips M, **Oliver-Bonet M**, Ko E, Rademaker A, Turek P, Martin RH.

    Reduced meiotic recombination on the XY bivalent is correlated with an increased incidence of sex chromosome aneuploidy in men with non-obstructive azoospermia. Mol Hum Reprod. 2008 Jul;14(7):399-404.

19. Obradors A, Fernández E, **Oliver-Bonet M**, Rius M, de la Fuente A, Wells D, Benet J, Navarro J

    Birth of a healthy boy after a double factor PGD in a couple carrying a genetic disease and at risk for aneuploidy: case report. Hum Reprod. 2008 Aug;23(8):1949-56.

20. Lian J, Yin Y, **Oliver-Bonet M**, Liehr T, Ko E, Turek P, Sun F, Martin RH

    Variation in crossover interference levels on individual chromosomes from human males. Hum Mol Genet. 2008 Sep 1;17(17):2583-94.

21. Sun F, Mikhaail-Philips M, **Oliver-Bonet M**, Ko E, Rademaker A, Turek P, Martin RH.
The relationship between meiotic recombination in human spermatocytes and aneuploidy in sperm. Hum Reprod. 2008 May 15. [Epub ahead of print]
22. Fernández L, Lapunzina P, Pajares IL, Palomares M, Martínez I, Fernández B, Quero J, García-Guereta L, García-Alix A, Burgueros M, Galán-Gómez E, Carbonell-Pérez JM, Pérez-Granero A, Torres-Juan L, **Heine-Suñer D**, Rosell J, Delicado A.
Unrelated chromosomal anomalies found in patients with suspected 22q11.2 deletion. Am J Med Genet A. 2008 May 1;146(9):1134-41.
23. Busquets X, MacFarlane NG, **Heine-Suñer D**, Morlá M, Torres-Juan L, Iglesias A, Lladó J, Sauleda J, Agustí AG.
Angiotensin-converting-enzyme gene polymorphisms, smoking and chronic obstructive pulmonary disease. Int J Chron Obstruct Pulmon Dis. 2007;2(3):329-34.
24. **Heine-Suñer D**, Torres-Juan L, Gómez C, Pérez-Granero A, Bernues M, Govea N, Rosell J.
Gene symbol: SRY.Hum Genet. 2007 Feb;120(6):909
25. Sun F, **Oliver-Bonet M**, Liehr T, Starke H, Ko E, Rademaker A, Martin RH.
Discontinuities and unsynapsed regions in meiotic chromosomes have a trans effect on meiotic recombination of some chromosomes in human males. Cytogenet Genome Res. 2007;119(1-2):27-32.
26. **Oliver-Bonet M**, Campillo M, Turek PJ, Ko E, Martin RH.
Analysis of replication protein A (RPA) in human spermatogenesis. Mol Hum Reprod. 2007 Dec;13(12):837-44
27. Torres-Juan L, **Rosell J**, Sánchez-de-la-Torre M, Fibla J, **Heine-Suñer D**.
Analysis of meiotic recombination in 22q11.2, a region that frequently undergoes deletions and duplications. BMC Med Genet. 2007 Apr 2;8:14.
28. Torres-Juan L, **Rosell J**, Morla M, Vidal-Pou C, García-Algas F, de la Fuente MA, Juan M, Tubau A, Bachiller D, Bernues M, Perez-Granero A, Govea N, Busquets X, **Heine-Suñer D**.
Mutations in TBX1 genocopy the 22q11.2 deletion and duplication syndromes: a new susceptibility factor for mental retardation. Eur J Hum Genet. 2007 Jun;15(6):658-63
29. Alonso MJ, **Heine-Suñer D**, Calvo M, Rosell J, Giménez J, Ramos MD, Telleria JJ, Palacio A, Estivill X, Casals T.
Spectrum of mutations in the CFTR gene in cystic fibrosis patients of Spanish ancestry. Ann Hum Genet. 2007 Mar;71(Pt 2):194-201.