

**ANTONI BORRELL, M.D., Ph.D.** (biography for ISPD Conference 08)

Senior Consultant in Obstetrics and Gynecology at the Hospital Clinic Barcelona and Associated Professor at the University of Barcelona Medical School. Trained in Fetal Medicine at Queen Charlotte's and Chelsea Hospital of London under supervision of Prof. C.H. Rodeck (1989), he has been on sabbatical studies in the Karolinska Institute Hospital (2005-2006), at Brigham & Women's Hospital, a Harvard Medical School affiliated hospital (2008-2009), and at Mount Sinai Hospital Toronto (2013). Interested in the fields of Fetal Medicine, Reproductive Genetics and Prenatal Diagnosis, particularly in ultrasound markers for fetal aneuploidy and early prenatal screening. Director of the International Society for Prenatal Diagnosis (ISPD), Chairman of the Fetal Ultrasound ISPD special interest group, and Deputy Chairman of the Educational Committee of the European Association for Perinatal Medicine (EAPM). President of the Catalan Chapter of Ultrasound in Obstetrics & Gynecology and advisor of the Catalan Health Department, has authored more than 100 peer reviewed international scientific publications.

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Dr. Antoni Borrell's interest is in the field of Fetal Medicine, particularly Prenatal Diagnosis and Screening of congenital anomalies, Fetal Ultrasound and Reproductive Genetics. An area of expertise is the ultrasound recognition of fetal syndromes, another is in prenatal genetic diagnosis and screening. He is Senior Consultant in Fetal Medicine at Hospital Clinic Barcelona, director of the prenatal diagnosis programme, and Associated Professor of the University of Barcelona. He has published extensively in the fields of interest. He is member of numerous scientific societies, and at present, he is the Chair of the First Trimester Quality Control Committee and advisor of the Catalan Health Department, and President of the Fetoscopy Working Group.

**ANTONI BORRELL, M.D., Ph.D.**

Graduated at the University of Barcelona Medical School in 1981.

Ph.D. degree at the University of Barcelona in 1995 ("Midtrimester ultrasound markers for aneuploidy").

Postgraduate Residency Training in Obstetrics and Gynecology at "Hospital Mutua de Terrassa" (Terrassa, Catalonia, Spain) (1983-1986).

Fellowship in Ultrasound and Prenatal Diagnosis in the Prenatal Diagnosis and Reproductive Counseling Unit of the University of Barcelona (Hospital Clinic) directed by Prof. A. Fortuny (1987-1988).

Fellowship in Fetal Medicine, advanced ultrasound and Doppler studies in the Institute of Obstetrics and Gynaecology, Royal Postgraduate Medical School, University of London, at Queen Charlotte's and Chelsea Hospital of London directed by Prof. C.H. Rodeck (1989).

Stage at the Harris Birthright Center for Fetal Medicine (King's College Hospital, University of London) (December 1989).

Sabbatical at the Molecular Medicine Center of Karolinska University Hospital, Stockholm, Sweden, under supervision of Dr. The-Hung Bui (2005-2006).

Sabbatical studies at Brigham & Women's Hospital affiliated at Harvard Medical School, Boston, MA (2007-2008).

Sabbatical at the Prenatal Diagnosis and Medical Genetics Program of the University of Toronto, Mount Sinai Hospital, Toronto, Ontario, Canada, supervised by David Chitayat (2013).

Consultant in Obstetrics and Gynecology in the Hospital Clinic – University of Barcelona Medical School ascribed to the Prenatal Diagnosis Unit (1990-1999). Regional Reference Centre for the Department of Health of Catalonia.

Head of the clinical area of Prenatal Diagnosis Unit of the Hospital Clinic – University of Barcelona Medical School (2000-2018).

Associated Professor in Obstetrics and Gynecology at the University of Barcelona Medical School (1997-2018).

Member of The Research Unit on Fetal-Maternal Medicine. IDIBAPS - Foundation for Biomedical Research. Hospital Clinic. University of Barcelona and CIBERER (Rare diseases research group).

Member of the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG)(from 1991), International Fetal Medicine and Surgery Society (IFMSS)(1994-2006), International Society for Prenatal Diagnosis and Therapy (ISPD)(from 1996) and Fetoscopy Working Group (from 2008).

Member of the Scientific Committee of the International Prenatal Screening Group (IPSG) formerly International Down's syndrome Screening Group (IDSSG) (2006-2012). Director of the International Society for Prenatal Diagnosis (2010-2014). Treasurer of the the International Society for Prenatal Diagnosis (2014-2018). Chairman of the Fetal Ultrasound ISPD special interest group (SIG) in 2012-2016 and Deputy-Chairman in 2010-2012.

Member of the Executive Boards of the Spanish Association for Prenatal Diagnosis (1998-2002) and President of the Ultrasound Chapter of the Catalan Society of Obstetrics and Gynecology (2011-2017).

Deputy Chairman del Educational Committee de la European Association for Perinatal Medicine (EAPM) (2012-2014).

Certificate of Ultrasound Level IV of The Section on Ultrasound of the Spanish Society for Obstetrics and Gynecology (2000).

Supervisor of 4 PhD projects in 2008, 2010, 2015 and 2017

Member of the Editorial Board of "Progresos en Diagnóstico Prenatal" (from 1997) and "Progresos en Obstetricia y Ginecologia" (from 2000).

Advisor of the Catalan Health Department in the writing of the protocol on Prenatal Diagnosis issued in 2009 and 2018.

Authored a number of international and national scientific publications and more than 160 peer-reviewed papers with impact factor

Selected peer-review papers in the last 5 years:

-Clinical application of midtrimester non-invasive fetal RHD genotyping and identification of RHD variants in a mixed-ethnic population. Prenat Diagn. 2013;33:173-178.

- Cumulative sum plots and retrospective parameters in first-trimester ductus venosus quality assurance. *Prenat Diagn.* 2013;33:384-390. doi: 10.1002/pd.4079.
- Cisterna magna width at 11-13 weeks in the detection of posterior fossa anomalies. *Ultrasound Obstet Gynecol.* 2013 May;41(5):515-20.
- First-trimester Down syndrome screening in renal-transplanted pregnant women: a model for adjusting the false-positives rates. *Prenat Diagn.* 2013;33:467-470
- First-trimester detection of major cardiac defects with the use of ductus venosus blood flow *Ultrasound Obstet Gynecol.* 2012;42:51-57
- Likelihood ratios to apply for nasal bone, ductus venosus and tricuspid flow at the 11-13 weeks' scan in Down syndrome screening. *Fetal Diagn Ther.* 2013;34(2):116-20
- Position statement from the Aneuploidy Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn.* 2013 Jul;33(7):622-9
- Birth defects in Medically Assisted Reproduction pregnancies in the city of Barcelona. *Prenat Diagn* 2014;34:327-334. doi: 10.1002/pd.4286.
- Is nuchal translucency a useful aneuploidy marker in fetuses with crown-rump length of 28-44 mm CRL? *Ultrasound Obstet Gynecol* 2014; 43: 520-524. doi: 10.1002/uog.13203.
- Maternal plasma DNA testing for aneuploidy in pregnancies achieved by assisted reproductive technologies. *Genet Med.* 2014;16:419-2. doi: 10.1038/gim.2013.149
- Antral follicle count as a marker of ovarian biological age to reflect the background risk of fetal aneuploidy. *Hum Reprod.* 2014 Jun;29(6):1337-43. doi: 10.1093/humrep/deu055.
- Nuchal translucency thickness in the prediction of unbalanced translocations. *Prenat Diagn.* 2014 Oct;34(10):982-5. doi: 10.1002/pd.4409. Epub 2014 Jun 11
- How to perform an amniocentesis. *Ultrasound Obstet Gynecol.* 2014 Dec;44(6):727-31. doi: 10.1002/uog.14680
- Array comparative genomic hybridization and fetal congenital heart defects: a systematic review and meta-analysis. *Ultrasound Obstet Gynecol.* 2015 Jan;45(1):27-35. doi: 10.1002/uog.14695.
- More chorionic villi obtained at a single center compared to previously published reports. *J Matern Fetal Neonatal Med.* 2015 Jan;28(1):46-8. doi: 10.3109/14767058.2014.900748. Epub 2014 Apr 9

-Further insights on diastolic dysfunction in first trimester trisomy 21 fetuses. *Ultrasound Obstet Gynecol.* 2015;45: 205-210. Epub 2014 Apr 4. doi: 10.1002/uog.13380. IF:3.14

-Role of ovarian reserve markers, antimüllerian hormone and antral follicle count, as aneuploidy markers in ongoing pregnancies and miscarriages. *Fertil Steril.* 2015 May;103(5):1221-1227.e2. doi: 10.1016/j.fertnstert.2015.02.022. Epub 2015 Mar 18.

-Position statement from the Chromosome Abnormality Screening Committee on behalf of the Board of the International Society for Prenatal Diagnosis. *Prenat Diagn.* 2015 Aug;35(8):725-34. doi: 10.1002/pd.4608. Epub 2015 Jun 4

-Genomic microarray in fetuses with increased nuchal translucency and normal karyotype – a systematic review and meta-analysis. *Ultrasound Obstet Gynecol.* 2015;46:650-658. doi: 10.1002/uog.14880. Epub 2015 Apr 20

-Transcervical chorionic villus sampling: a practical guide. *J Matern Fetal Neonatal Med.* 2016 Apr;29(8):1244-51. doi: 10.3109/14767058.2015.1043261. Epub 2015 Jun 11

-Intertwin crown-rump length discordance in the prediction of fetal anomalies, fetal loss and adverse perinatal outcome. *J Matern Fetal Neonatal Med.* 2016 Sep;29(17):2883-8. doi: 10.3109/14767058.2015.1107901. Epub 2015 Nov 23.

-Effectiveness of ovarian age as the background risk for aneuploidy screening in an unselected pregnant population. *Reprod Biomed Online.* 2016 Oct;33(4):500-505. doi: 10.1016/j.rbmo.2016.07.006.

- Crown-rump length audit plots with the use of operator-specific PAPP-A and  $\beta$ -hCG median MoM. *Prenat Diagn.* 2017 Mar;37(3):229-234. doi: 10.1002/pd.4996. Epub 2017 Jan 25.

- Heterotrisonomy recurrence risk: a practical maternal age-dependent approach for excess trisomy 21 risk calculation after a previous autosomal trisomy. *J Matern Fetal Neonatal Med.* 2017 Jul;30(13):1613-1615. doi: 10.1080/14767058.2016.1219990. Epub 2016 Sep

- Genomic Microarray in Fetuses with Early Growth Restriction: A Multicenter Study. *Fetal Diagn Ther.* 2017;42(3):174-180. doi: 10.1159/000452217. Epub 2016 Nov 2.

- Overview of Chromosome Abnormalities in First Trimester Miscarriages: A Series of 1,011 Consecutive Chorionic Villi Sample Karyotypes.

Cytogenet Genome Res. 2017;152(2):81-89. doi: 10.1159/000477707. Epub 2017 Jun 30.

-Chromosomal Microarray Analysis in Fetuses with Growth Restriction and Normal Karyotype: A Systematic Review and Meta-Analysis. Fetal Diagn Ther. 2018;44(1):1-9. doi: 10.1159/000479506. Epub 2017 Sep 9.

-Parental Origin of the Retained X Chromosome in Monosomy X Miscarriages and Ongoing Pregnancies. Fetal Diagn Ther. 2017 Oct 5. doi: 10.1159/000480499. [Epub ahead of print]

-Added value of chromosomal microarray analysis over karyotyping in early pregnancy loss: systematic review and meta-analysis. Ultrasound Obstet Gynecol. 2018 Apr;51(4):453-462. doi: 10.1002/uog.18929. Review

-Counting ovarian antral follicles by ultrasound: a practical guide. Ultrasound Obstet Gynecol. 2018 Jan;51(1):10-20. doi: 10.1002/uog.18945.

Editorial commentaries in the last 5 years:

-Miscarriage in contemporary maternal-fetal medicine: targeting clinical dilemmas. Ultrasound Obstet Gynecol. 2013; 42:491-497 Feb 22. doi: 10.1002/uog.12442

-Cell-free DNA testing: inadequate implementation of an outstanding technique. Ultrasound Obstet Gynecol. 2015 May;45(5):508-11. doi: 10.1002/uog.14795

-A new comprehensive paradigm for Prenatal Diagnosis. Ultrasound Obstet Gynecol. 2018 Feb 26. doi: 10.1002/uog.19035. [Epub ahead of print] PMID:29484739