Carlos Venegas-Vega
Hospital General de Mexico
Dr. Balmis 148
6720 Mexico DF, Mexico

E-Mail: cavenevega@gmail.com

ESHG 2016 - INFORMATION ON ABSTRACT ACCEPTANCE
ELECTRONIC POSTER PRESENTATION

Dear colleague,

On behalf of the Scientific Programme Committee of the European Conference of Human Genetics 2016 taking place in Barcelona, Spain from May 21 to May 24, 2016 we are pleased to inform you that the abstract entitled

'Microdeletion of 10q21.3-q22.1 including CTNNA3 in a boy with intellectual disability, mucocutaneous pigmentation, and juvenile colon polyposis'
(Control No. 2016-A-2526-ESHG)
has been accepted for **Electronic Poster Presentation**.

Schedule

Electronic Posters will be on display in the Poster Area and can be accessed at during exhibition opening hours from **Saturday, May 21 (09:30 hrs)** to **Monday, May 23 (17:45 hrs)** by all participants. Please upload your presentation upon your arrival in the **Preview Centre** from **Friday, May 20 from 15:00 hrs onwards** (during conference times).

Format

Please create a short presentation (either in PowerPoint or PDF) with a maximum of 4 slides with the most important contents of your study. This is to be uploaded in the Preview Centre.
File format: .pdf or .ppt/.pptx
Presentation format: 16:9, portrait(!)

The presentations will be displayed on several 42-inch touch screens in the Poster Area in portrait format.
The system will automatically generate buttons to navigate through the slides.

The **website** gives more details on your presentation format (menu item "Information - Info for Presenters – Electronic Poster").

Please note that due to new EACCME regulations, **authors are requested to disclose possible conflicts of interest** on the electronic poster.

All details on your poster presentation (e.g. E-Poster number) will be sent in the first week of May 2016.

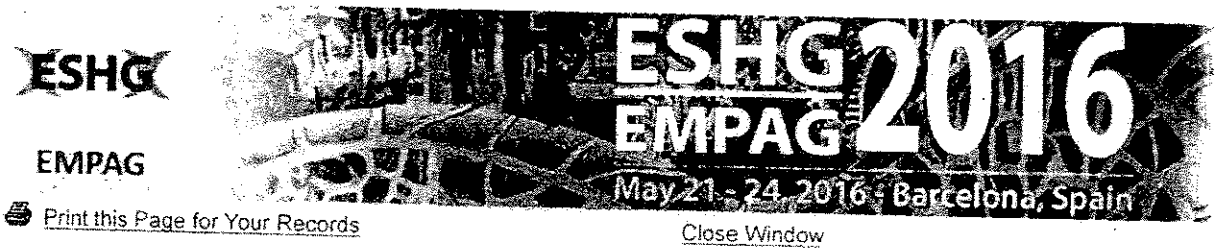
Please remember that the deadline for **reduced registration fee is March 31, 2016.** We also advise you to book your **hotel accommodation in Barcelona** as soon as you can. Registration and hotel booking is possible on the website: www.eshg.org/eshg2016.

We look forward to welcoming you in Barcelona in May!

With kind regards,

Brunhilde Wirth Joris Veltman
Co-Chairs of the Scientific Programme Committee

Contact Organising Office
ESHG 2016
c/o Vienna Medical Academy
Alser Str. 4
1090 Vienna, Austria
t: 0043 (0) 1 405 13 83 11
f: 0043 (0) 1 407 82 74
e: conference@eshg.org
w: www.eshg.org/eshg2016



Control/Tracking Number: 2016-A-2526-ESHG
Activity: ESHG Abstract
Current Date/Time: 2/15/2016 11:33:28 AM

Microdeletion of 10q21.3-q22.1 including CTNNA3 in a boy with intellectual disability, mucocutaneous pigmentation, and juvenile colon polyposis

Author Block: C. Venegas-Vega[1,2], M. Guardado[1], J. Berumen[1,2];
[1]Hospital General de Mexico, Mexico DF, Mexico, [2]Facultad de Medicina, Universidad Nacional Autónoma de México, UNAM, Cdmx, Mexico.

Abstract:
Introduction: We described the case of a 9-year-old boy with a de novo 10q21.3-q22.1 microdeletion, intellectual disability (ID), attention-deficit hyperactivity disorder, mucocutaneous pigmentation, and juvenile colon polyposis (JCP). Methods and Results: Cytogenetic analyses in the proband and her parents was normal; Chromosomal Microarray Analysis (CMA) by CytoScan Optima in the patient revealed a deletion of about 4.41 Mb ([hg19] chr10:68,299,927-72,715,960), while CMA of parental DNA was normal. Although no similar size deletions were reported in DECIPHER database, there are several patients with much smaller pure deletions (86.05 to 378.77 Kb) that clustered at CTNNA3 gene (chr10:68,165,545-69,222,108). The most remarkable common feature of these cases was intellectual disability, global developmental delay and autism spectrum disorder. Moreover, the analysis of the gene content of the deleted region showed 40 MIM genes; of which only four have been implicated in autosomal dominant diseases. Our patient does not exhibit any clinical data of those conditions. In order to identify that genes could be involved in the phenotype, we analyzed physiologic roles, functional interactions, and also which those are dosage-sensitive. The results pointed to CTNNA3 (607667), LRRTM3 (610869) and SIRT1 (604479); all located in 10q21.3, as best candidates. Thus, these three genes may either individually or in combination be responsible for ID with JCP seen in our patient. Conclusions: To the best knowledge, this is the first report of a de novo 10q21.3-q22.1 deletion. This case suggests a novel genomic disorder at 10q21.3 region associated to ID with JCP.

Author Disclosure Information: C. Venegas-Vega: None. M. Guardado: None. J. Berumen: None.
Topic (Complete): 08. Intellectual Disability
Keyword (Complete): 10q21.3 deletion ; CTNN3A ; intellectual disability with colon polyposis
Presentation Preference (Complete): Poster only
Awards - Fellowships (Complete):
YoungScientistAward: No
Application for Fellowship: No
Application for Fellowship: No
Application for Fellowship: No

Status: Complete

Invitation to the Conference

Dear Colleagues and Friends,

The last time the ESHG was held in Spain was in 2008 in Barcelona and it is about time to act again as hosts for a new edition of our scientific meeting. It gives me great pleasure, to invite you on behalf of the Spanish Association of Human Genetics (AEGH) to Barcelona for the 2016 Conference from May 21 – 24.

Barcelona is a city that needs no much of an introduction, it is Catalonia's largest city renowned for its culture and art and offers Mediterranean life-style.
Catalonia, and Barcelona itself have managed to be the source of much of the country's creativity, including gastronomic culture, art and architecture. But although Barcelona's cultural figureheads (Picasso, Dali, Gaudi) continue to hold plenty of appeal, the city has more to offer than the creations of these famous residents. From the medieval quarter via the elegant 19th century Eixample to the regenerated waterfront, the city rewards the curious pedestrian, its streets awash with beauties, curiosities and diversions. Indeed, the very urban fabric of Barcelona is deliriously vital: grand with medieval design, playful with impish street art and ablaze with Modernista colours and furbelows.

Barcelona is also notable for being an international hub for science. The city has a very rich ecosystem of prestigious scientific institutes and scientists, many of them working in Human Genetics and Bioinformatics.

The 2016 conference will provide, among other elements, the latest in the field of applied genomics and, hopefully, some real pathways to translate the genomic findings into clinical genetics applications. It will be for sure inspirational and an event not to be missed.

See you in Barcelona in 2016!

Juan C. Cigudosa, Local Host
President, Spanish Society of Human Genetics

The European Society of Human Genetics promotes research in basic and applied human and medical genetics and facilitates contact between all persons who share these aims.

General Information

This international conference (now in its 49th year) is a forum for all workers in human and medical genetics to review advances and develop research collaborations. The conference has become one of the premier events in the field of human genetics with over 3,000 delegates, more than 200 oral presentations, 18 workshops and 8 educational sessions. The ESHG conference is where the latest developments in human genetics are discussed and where professionals from all parts of human genetics meet.

Programme	• Invited Plenary lectures and Symposia • ESHG Award and Mendel lectures • "Educational Track" throughout the meeting • Workshops • Concurrent sessions of submitted abstracts • Poster presentations of submitted abstracts • Young Scientist and Poster Awards • Conference Fellowships for young researchers from Central and Eastern Europe, as well as Fellowships for National Societies • Corporate Satellites • Over 150 exhibitors from all over the world
Further details	The website **www.eshg.org/eshg2016** will open in October 2015
Abstract submission	Online abstract submission via www.eshg.org/eshg2016 **Closing date: Friday February 12, 2016**
Scientific & Administrative Conference Secretariat	ESHG 2016 c/o Vienna Medical Academy Alser Strasse 4, 1090, Vienna, Austria Tel: +43 1 405 13 83 11 Email: conference@eshg.org
Exhibition, Sponsoring, Corporate Satellites	Rose International P.O.Box 93260, 2509 AG The Hague The Netherlands Tel: +31 70 383 8901 Email: eshg@rose-international.com

Further information on programme, registration and abstract submission:

www.eshg.org/eshg2016

Centre de Convencions Internacional de Barcelona

European Human Genetics Conference

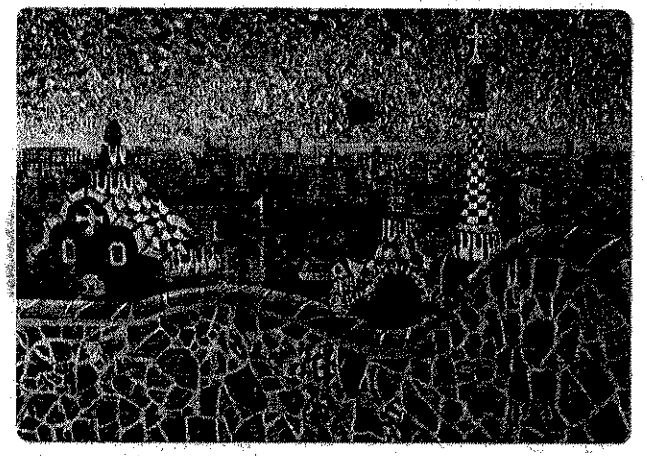
ESHG EMPAG 2016

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

Barcelona, Spain, May 21 - 24, 2016

In conjunction with the
European Meeting on Psychosocial Aspects of Genetics

EMPAG
EUROPEAN MEETING ON PSYCHOSOCIAL ASPECTS OF GENETICS



www.eshg.org/eshg2016

Preliminary Schedule ESHG 2016

The preliminary programme is subject to alteration. Please consult the website as of October 2015.
New! On site registration opens on Friday, May 20, 15:00 - 18:00 hrs

Saturday, May 21

Time	
08.30	*Registration opens*
10.15	
10.30	**Educational Sessions** ES1. Novel genome sequencing technologies ES2. Genetic Privacy & Data Sharing (with EMPAG) ES3. Generation & use of induced pluripotent stem cells ES4. Peroxisomal disorders - still a need for metabolic assays? **Workshops** WS01 Computational Methods for Human Genetics WS02. A case that changed my life as a geneticist WS03. Career development & funding opportunities for young investigators WS04. Pharmaco-genomics
12.00	*Lunch - Posters - Exhibition*
14.00	**Welcoming Addresses** **Opening**
14.30	**Opening Plenary Session 1**
16.00	*Coffee - Posters - Exhibition*
16.30	**Plenary Session 2** **What's new? Highlights**
18.00	*Coffee - Posters - Exhibition*
18.30	**Concurrent Sessions C01 - C05**
20.00	*Networking Mixer*

Corporate Satellites

Sunday, May 22

Time	
08.30	**Concurrent Symposia S01-S04** S01. The future lies in uncertainty S02. Understanding functional effects of genomic variants S03. Hereditary cancer S04. From families to populations and back
	Educational Session ES5. Ciliopathies
10.00	*Coffee - Posters - Exhibition*
10.15	**Poster Viewing with Authors - Group 1**
11.15	*Lunch - Posters - Exhibition*
13.00	**Concurrent Sessions C07 - C12**
14.30	*Vitamin Break - Posters - Exhibition*
15.00	**Workshops W05-W11** WS05. NGS in the Clinic WS06. Dysmorphology 1 WS07. CNV Interpretation & Classification WS08. Pre-conception carrier testing WS09. How to get published WS10. Genome Browser UCSC WS11. Gene regulation
16.30	*Coffee - Posters - Exhibition*
16.45	**Poster Viewing with Authors - Group 2**
17.45	**Concurrent Symposia S05 - S08** S05. Circulating Cell-Free Nucleic Acids S06. Behavioural disorders S07. Long distance regulation of transcription & translation S08. Sensory disorders
	Educational Session ES6. Clinical interpretation of genetic variants
19.15	*ESHG Membership Meeting*
20.00	

Corporate Satellites

Monday, May 23

Time	
08.30	**Concurrent Symposia S09-S12** S09. Debating germline genome editing S10. Brain genetics S11. Human genomics: transcriptome, proteome & interactome S12. SeX and the city
	Educational Session ES7. Cleaning the noise from Big Data
10.00	*Coffee - Posters - Exhibition*
10.15	**Poster Viewing with Authors - Group 3**
11.15	*Lunch - Posters - Exhibition*
13.00	**Concurrent Sessions C13 - C18**
14.30	*Vitamin Break - Posters - Exhibition*
15.00	**Workshops W12-W18** WS12. Reproductive genetics WS13. Dysmorphology 2 WS14. CRISPR CAS9 tips and tricks WS15. Rare Diseases and clinical trials WS16. Clinical Cancer Genetics WS17. ENGEMBL WS18. The clinic: From face-to-face to interface
16.30	*Coffee - Posters - Exhibition*
16.45	**Poster Viewing with Authors - Group 4**
17.45	**Concurrent Symposia S13 - S16** S13. Reproductive genetics and genomics S14. Epigenetic reprogramming in disease S15. Therapy in rare diseases S16. Cardiovascular genetics - with ESC
	Educational Session ES8. Clinical interpretation of genetic variants
19.15	Corporate Satellites
20.00	*Networking Event*

Corporate Satellites

Tuesday, May 24

Time	
09.00	**Plenary Session 3** PL3. My vision on Genomic Medicine
10.30	*Coffee Break*
11.00	**Concurrent Sessions C19 - C24**
12.30	*Lunch Break*
13.30	**Plenary Session 4** **Mendel Lecture** **Plenary Session 5** **ESHG Award Lecture** **EJHG Nature Awards** **Young Investigator Awards** **ESHG Poster Awards** **Closing**
14.15	
15.45	*End of Meeting*

Plenary Sessions | Concurrent Symposia | Concurrent Sessions | Educational Sessions | Workshops | Corporate Satellites

Deadline for submitting abstracts: Friday, February 12, 2016 www.eshg.org/eshg2016

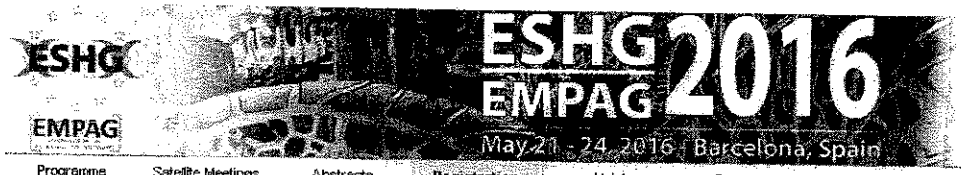
Committees

ESHG Scientific Programme Committee

Joris Veltman, *Co-Chair*, NL
Brunhilde Wirth, *Co-Chair*, DE
Juan Cigudosa, *Local Host*, ES
Yanick Crow, FR
Helene Dollfus, FR
Francesca Forzano, *Observer*, IT
Brunella Franco, IT
Lude Franke, NL
Maurizio Genuardi, IT
Daniel Grinberg, ES
Gunnar Houge, *ex officio*, NO
Erik Iwarsson, SE
Xavier Jeunematire, FR
Pablo Lapunzina, ES
Conxi Lazaro, ES
José Carlos Machado, PT
William Newman, UK
Minna Nyström, FI
Lucy Raymond, UK
Elisabella Razzaboni, IT
Alexandre Reymond, CH
Sam Riedijk, NL
Samuli Ripatti, FI
Peter Robinson, DE
Maria Jesus Sobrido, ES
Eduardo Tizzano, ES
Kristel van Steen, BE
Joris Vermeesch, BE
Karin Writzl, *ex officio*, SI

ESHG Annual Meetings Committee

Andrew Read, UK, AMC Chair
Feliciano Ramos, ES, ESHG President
Helena Kääriäinen, FI, Vice-President
Gunnar Houge, NO, Secretary General
Karin Writzl, SI , Deputy Sec. General
Jerome del Picchia, AT, Executive Officer
Jantie de Roos, NL, Advisor
Kristina Libova, AT, Advisor
Mirjam Liebelhör, AT, Advisor



ESHG 2016

Official Events
General Terms and Conditions

Registration

Registration Fees

PAYMENT RECEIVED:	*before March 31 (reduced rate)*	*from April 1 to May 1 (regular rate)*	*after May 1 and on site*	*Day Tickets on site*
Participants ESHG Members	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Participants Non-Members	EUR 450.-	EUR 550.-	EUR 600.-	EUR 200.-
Postgrad. Trainees ESHG Members[1]	EUR 200.-	EUR 300.-	EUR 350.-	EUR 125.-
Postgrad. Trainees Non-Members[1]	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Counsellors/Gen.Nurses ESHG Members[2]	EUR 200.-	EUR 300.-	EUR 350.-	EUR 125.-
Counsellors/Gen.Nurses Non-Members[2]	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Students[3]	EUR 100.-	EUR 150.-	EUR 200.-	EUR 100.-
Lunch bags/boxes per day	EUR 16.-	EUR 16.-	N/A	N/A

	Participants	**Students/Postgraduate Trainees**
Networking Evening at own expense	EUR 55.-	EUR 35.-

[1]Applies to *MSc./PhD students*. Please provide a confirmation signed by the head of department **at the moment of your registration** by fax to +43 1 407 82 74 or via email to **conference(at)eshg.org**. **Confirmations handed in at a later stage cannot be considered**.

Conversor de divisas XE

Conversor | Tipos de cambio | Análisis | Información

600,00 EUR = 11.882,56 MXN

Euro ↔ Peso mexicano
1 EUR = 19,8043 MXN 1 MXN = 0,0504942 EUR

Consultar tabla

Volver a convertir

Tipos de cambio intermedio: 2016-04-15 18:43 GMT

License XE Currency Rates. Click here!!

Carlos Venegas Vega
Hospital General de Mexico
Dr. Balmis 148
6720 Mexico DF, Mexico

E-Mail: cavenevega@gmail.com

ESHG 2016 - INFORMATION ON ABSTRACT ACCEPTANCE
ELECTRONIC POSTER PRESENTATION

Dear colleague,

On behalf of the Scientific Programme Committee of the European Conference of Human Genetics 2016 taking place in Barcelona, Spain from **May 21 to May 24, 2016** we are pleased to inform you that the abstract entitled

'Microdeletion of 10q21.3-q22.1 including CTNNA3 in a boy with intellectual disability, mucocutaneous pigmentation, and juvenile colon polyposis'
(Control No. 2016-A-2526-ESHG)

has been accepted for **Electronic Poster Presentation**.

Schedule

Electronic Posters will be on display in the Poster Area and can be accessed at during exhibition opening hours from **Saturday, May 21 (09:30 hrs)** to **Monday, May 23 (17:45 hrs)** by all participants. Please upload your presentation upon your arrival in the **Preview Centre** from **Friday, May 20 from 15:00 hrs onwards** (during conference times).

Format

Please create a short presentation (either in PowerPoint or PDF) with a maximum of 4 slides with the most important contents of your study. This is to be uploaded in the Preview Centre.
File format: .pdf or .ppt/.pptx
Presentation format: 16:9, portrait(!)

The presentations will be displayed on several 42-inch touch screens in the Poster Area in portrait format.
The system will automatically generate buttons to navigate through the slides.

The **website** gives more details on your presentation format (menu item "Information - Info for Presenters – Electronic Poster").

Please note that due to new EACCME regulations, **authors are requested to disclose possible conflicts of interest** on the electronic poster.

All details on your poster presentation (e.g. E-Poster number) will be sent in the first week of May 2016.

Please remember that the deadline for **reduced registration fee is March 31, 2016.** We also advise you to book your **hotel accommodation in Barcelona** as soon as you can. Registration and hotel booking is possible on the website: www.eshg.org/eshg2016.

We look forward to welcoming you in Barcelona in May!

With kind regards,

Brunhilde Wirth Joris Veltman
Co-Chairs of the Scientific Programme Committee

Contact Organising Office | **ESHG 2016** c/o Vienna Medical Academy Alser Str. 4 1090 Vienna, Austria | t: 0043 (0) 1 405 13 83 11 f: 0043 (0) 1 407 82 74 e: conference@eshg.org w: www.eshg.org/eshg2016