TWENTY-SEVENTH EUROPEAN MEETING ON DYSMORPHOLOGY

7 – 9 SEPTEMBER 2016
LE BISCHENBERG

27th EUROPEAN MEETING ON DYSMORPHOLOGY

GENERAL PROGRAM

WEDNESDAY 7th SEPTEMBER

5 p.m. to 7.30 p.m.		Registration
7.30 p.m. to 8.30 p.m.		Welcome reception
8.30 p.m.	Dinner	
9.30 p.m.		Unknown [KD1]

THURSDAY 8th SEPTEMBER

8.15 a.m.		Opening address
8.30 a.m. to 1.00 p.m.		First, second and third sessions
1.00 p.m.	Lunch	
2.30 p.m. to 7.00 p.m.		Third session
8.00 p.m.	Dinner	
9.00 p.m. to 11.00 p.m.		Unknown

FRIDAY 9th SEPTEMBER

8.30 a.m. to 1.00 p.m.		Fourth and fifth sessions
1.00 p.m.	Lunch	
2.30 p.m. to 6.00 p.m.		Sixth and seventh sessions
7.30 p.m.	Dinner	

SATURDAY 10th SEPTEMBER

Breakfast – Departure

SCIENTIFIC PROGRAM

Note: This program is tentative and may be modified.

WEDNESDAY 7th SEPTEMBER

9.30 UNKNOWN SESSION
Chair: VERLOES A.

L. BASEL VANAGAITE, K. DEVRIENDT AND NICOLE FLEISCHER
Workshop: Improving dysmorphology skills of new syndromes with computer aided analysis

THURSDAY 8th SEPTEMBER

08.30 Opening address: STUMPEL C.

08.45-10.00 FIRST SESSION: Craniofacial malformations and dysmorphology
Chair: STOLL C.

08.45 C. STOLL, Y. ALEMBIK, B. DOTT AND M.-P. ROTH
Associated anomalies in cases with anotia and microtia

09.00 D. LACOMBE, M. BERENGUER, E. LOPEZ, A. TINGAUD-SEQUEIRA, S. MARLIN, A. TOUTAIN, F. DENOYELLE, A. PICARD, S. CHARRON, G. MATHIEU, H. DE BELVALET, B. ARVEILER, P.J. BABIN AND C. ROORYCK
Mutations in *MYT1*, encoding the myelin transcription factor 1, are a rare cause of Goldenhar syndrome within the RA signaling pathway

09.15 A. VERLOES, D. HAYE, C. BAUMANN, A. TOUTAIN, D. BONNEAU, D. LEHALLE, P. CALLIER, S. DRUNAT, I. LUND, E. BHOJ, P. BOGAARD, S. LEENSKJOLD, E. ZACKAI, D. LI, H. HAKONARSON, M. PETERSEN AND I. NIELSEN
Phenotypic spectrum of *SPECC1L* mutations encompasses Teebi hypertelorism syndrome but not Opitz BBBG syndrome: report of new families and critical review of the literature

09.30 E. BRISCHOUX-BOUCHER, C. COLLET, C. CZORNY AND L. VAN MALDERGEM
Crouzon-like craniosysnostosis in two siblings harboring *IL11RA* composite heterozygous mutations

09.45 J. ROOSENBOOM, I. SAEY, A. VERDONCK, V. VANDER POORTEN, T. DORMAAR, J. SCHOENAERS, K. DEVRIENDT, H. PEETERS, P. CLAES AND G. HENS
Facial morphology associated with isolated cleft palate

10.00-11.00 SECOND SESSION: Skeletal dysplasias

10.00 A. MATULEVIČIENĖ, L. AMBROZAITYTĖ, R. MATULEVIČIŪTĖ, A.LIUBŠYS, A. UTKUS AND V. KUČINSKAS
A patient with Raine syndrome due to novel mutations in *fam20c* gene

10.15 A. BAYAT, B. KERR AND S. DOUZGOU
The evolving cranio-facial phenotype and a bifid uvula in a patient with Sensenbrenner syndrome caused by *IFT140* compound heterozygous mutations

10.30 M. JEANNE, S. ALIROL, N. CHELLOUG, L. DRUART, E. TERRENOIRE, N. RONCE, M. RAYNAUD AND A. TOUTAIN
A new case of X;Y translocation further supports the existence of a growth control locus on the Y chromosome

10.45 K. STEINDL, P. JOSE, L. GOGOLL AND A. RAUCH
if not Albright hereditary osteodystrophy what is it? Biallelic variants in the PRMT7 gene cause a novel syndrome with a distinct Albright-like phenotype

11.00-11.30 *Coffee Break*

11.30-12.30 THIRD SESSION: Clinical and genetic delineation of known syndromes (part 1)
Chair: LACOMBE D. – RAUCH A.

11.30 S. PASSEMARD, K. IZUMI, M. BRETT, E. NISHI, S. DRUNAT, E.-S. TAN, K. FUJIKI, S. LEBON, B. CHAM, K. MASUDA, M. ARAKAWA, A. JACQUINET, Y. YAMAZUMI, S.-T. CHEN, A. VERLOES, Y. OKADA, Y. KATOU, T. NAKAMURA, T. AKIYAMA, P. GRESSENS, R. FOO, E.-C. TAN, V. EL GHOUZZI AND K. SHIRAHIGE
ARCN1 mutations in a dominant form of microcephalic dwarfism

11.45 M. ZENKER, D. SCHANZE, J. RAO, W. TAN, D. BRAUN, D. SCHAPIRO, F. HILDEBRANDT and the International GAMOS Consortium
Nephrotic syndrome with associated brain anomalies – new lessons on the heterogeneity of Galloway-Mowat syndrome

12.00 C. FAUTH, G. KROPSHOFER, A. SCHOSSIG, L. PÖLSLER, B. MEISTER and J. ZSCHOCKE
Prolidase deficiency - a rare autosomal recessive disorder associated with intellectual disability, short stature and facial dysmorphism case report of a 13-year-old boy

12.15 I. IVANOVSKI, S. ROSATO, I. MAINI, M. MUSSINI, K. STEINDL, M. ZWEIER, L. GARAVELLI AND A. RAUCH
A new case of Helsmoortel-van der Aa syndrome (ADNP-related intellectual disability and autism spectrum disorder)

12.30 L. GOGOLL, P. JOSET, K. STEINDL AND A. RAUCH
A new case of Ogden syndrome: a review on the clinical course and the natural history of a rare lethal X-linked recessive disorder

AFTERNOON

14.30-16.00 THIRD SESSION: Clinical and genetic delineation of known syndromes (part 2)
Chair: BIJLSMA E. – BONATI M.T.

14.30 I. MEERSCHAUT, S. DE CONINCK, S. GARCIA MIÑAUR, J.C. OOSTERWIJK, R. IGBOKWE, M. SURI, A. BAYAT, G. JONES, C.I. DALI, S. LYNCH, E. BLAIR, A. COLLINS, V. LAITINEN, E. THOMAS, A. MALE, I. STOLTEDIJKSTRA, K. KEYMOLEN, L. CHERYL, D. YADAV, F. MCKENZIE, S. BERLAND, P. WILLEMS, A. TOPA, F. PETIT, A. DESTREE, J. DE BACKER, P.J. COUCKE, A. DE PAEPE, S. SYMOENS AND B. CALLEWAERT
Congenital contractural arachnodactyly: delineation of clinical diagnostic criteria

14.45 L. GARAVELLI, I. IVANOVSKI, S.G. CARAFFI, D. SANTODIROCCO, M. POLLAZZON, D.M. CORDELLI, E. ABDALLA, P. ACCORSI, M.P. ADAM, P. AJMONE, M. BADURA-STRONKA, C. BALDO, A. BAYAT, E. BELLIGNI, S. BIGONI, F. BONVICINI, J. BRECKPOT, B. CALLEWAERT, G. COCCHI, G. CUTURILO, D. DE BRASI, K. DEVRIENDT, M.B. DINULOS, O. DJURIC, T. DUELUND HJORTSHØJ, R. EPIFANIO AND F. FARAVELLI, A. FIUMARA, D. FORMISANO, L. GIORDANO, M. GRASSO, S. GRØNBORG, A. IODICE, L. IUGHETTI, V. KUBUROVIC, A. KUTKOWSKA-KAZMIERCZAK, D. LACOMBE, C. LO RIZZO, A. LUCHETTI, B. MALBORA, I. MAMMI, F. MARI, S. MOUTTON, R. MØLLER, P. MUSCHKE, E. OBERSZTYN, C. PANTALEONI, A. PELLICCIARI, M.A. PISANTI, M.L. POCH OLIVE, F. RAVIGLIONE, F. RIVIERI, S. SAVASTA, G. SCARANO, I. SCHANZE, A. SELICORNI, M. SILENGO, R. SMIGIEL, L. SPACCINI, G. SORGE, K. SZCZALUBA, L. TARANI, L. GONZAGA TONE, A. TOUTAIN, A. TRIMOUILLE, E. TERCI VALERA, S. SCHRIER VERGANO, N. ZANOTTA, M. ZENKER, M. ZOLLINO, C. ZWEIER, W.B. DOBYNS AND A.R. PACIORKOWSKI
Clinical findings in Mowat-Wilson syndrome: a study of 80 patients

15.00 A. KUECHLER, B. ALBRECHT, E. LEGIUS, N.C. BRAMSWIG, H.-J. LÜDECKE, T.M. STROM, D. WIECZOREK, M. ZENKER AND I. WIELAND
An adult female patient with ocular dermoid, calcifying meningeomas, hyperostosis of the skull,facial, alveolar and phalangeal exostoses and skin changes - a long way to the right diagnosis

15.15 K. KEYMOLEN, L. DE MEIRLEIR, M. DE RADEMAEKER, D. DANEELS, B. CALJON, S. VAN DOOREN, D. CROES AND E. LEGIUS
Overgrowth and intellectual disability: two more patients with DNMT3A mutations

15.30 A. VINCENT, V. GATINOIS, A. FAVREAU AND A. TOUTAIN
Pituitary stalk interruption syndrome in Kabuki syndrome: a case report and review of the literature

15.45 C. STUMPEL, J. NICOLAI, S. STEGMANN, K. OBERNDORFF, C. GEUSGENS, I. KEULARTS-KÖRVER AND E. RUBIO-GOZALBO
A girl with two mutations in PIGN: a challenging diagnosis

16..00-16.30 *Coffee Break*

16.30-17.30 THIRD SESSION: Clinical and genetic delineation of known syndromes (part 3)
Chair: STUMPEL C.- KOHLHASE J.

16.30 I. SLEGERS, M. LEYDER, K. VAN BERKEL, S.A. KLUIJFHOUT, M. CANNIE, C. STAESSEN, G. LEEMANS, A. JANSEN, M. DE RADEMAEKER, B. DIMITROV, M. BONDUELLE AND K. KEYMOLEN
Fetal (prenatal) features of PITT Hopkins syndrome - case report

16.45 D. BARTHOLDI, D. DOECKER, L. VOGEL, J. SCHUBERT, E.M. GOLDMANN AND S. BISKUP
Identification of a heterozygous *de novo* variant in the metabotropic glutamate receptor type 5 gene (*GRM5*) in a patient with non-syndromic intellectual disability – a novel rare cause of developmental disabilities?

17.00 I.K. NIELSEN, I.S. PEDERSEN, V.Q. LE, A. ERNST AND J.R. OSTERGAARD
Recurrence of *MEF2C mutation in siblings with unusual communicative skills*

17.15 D. MITTER, M. PRINGSHEIM, M. KAULISCH, K. PLÜMACHER, S. SCHRÖDER, D. MALZAHN, B. ZIRN, K. BROCKMANN AND FOXG1-STUDY GROUP
FOXG1 syndrome: genotype-phenotype correlation in 84 patients with a *FOXG1* point mutation

17.30 KEY-NOTE LECTURE
A. BLOCH-ZUPAN
Amelogenesis imperfecta: lessons from NGS

21.00-23.00 UNKNOWN
Chair: VERLOES A. – DEVRIENDT K.

K. AVELA, K. AALTONEN AND K. AITTOMÄKI
Formal unknown: An unknown case with growth hormone deficiency and dysmorphic features

A. BOUMAN and P. LAKEMAN
A 36-year-old female with a Treacher Collins-like phenotype plus additional features

FRIDAY 9th SEPTEMBER

08.45-11.00 FOURTH SESSION: X-linked intellectual disability
Chair: VERLOES A. MIDRO A.

08.45 S. MOORTGAT, D. LEDERER, M. DEPREZ, M. BUZATU, S. BOULANGER, V. BENOIT, S. MARY AND I. MAYSTADT
Expanding the phenotype of *OPHN1* mutations: three unrelated families with intellectual disability and absence of cerebellar hypoplasia

09.00 S. MOORTGAT, J. DESIR, V. BENOIT, S. BOULANGER, H. PENDEVILLE, M.-C. NASSOGNE, D. LEDERER AND I. MAYSTADT
Two Novel EIF2S3 mutations associated with X-linked syndromic intellectual disability with severe microcephaly, growth retardation, and epilepsy

09.15 A. BOUMAN, M. ALDERS, L. VAN LEEUWEN, N. THUIJS AND M. VAN MAARLE
OFD1 in males: congenital heart defect can be included in its phenotypic spectrum

09.30 M. DE RADEMAEKER, L. DEMEIRLEIR, A.C. HOUTMAN, K. STOUFFS, B. CALJON, S. VAN DOOREN, D. CROES AND K. KEYMOLEN
A BCOR variant in a male

09.45 N. DIKOW, M. GRANZOW, L.M. GRAUL-NEUMANN, S. KARCH, K. HINDERHOFER, N. PARAMASIVAM, L.-J. BEHL, L. KAUFMANN, C. FISCHER, C. EVERS, M. SCHLESNER, R. EILS, G. BORCK, C. ZWEIER, C.R. BARTRAM, J.C. CAREY AND U. MOOG
Phenotypic overlap in girls with *DDX3X* mutations and Toriello-Carey syndrome

10.00 O.M. VANAKKER, B. LAPAUW AND A. GEERTS
Two novel patients with an Xq28 duplication excluding *FMR1* and *MECP2*

10.15 J. KÖTTING, J. BEYGO, H.-J. LÜDECKE, K. BUITING AND A. KÜCHLER
5-year old boy with intellectual disability, dysmorphic facial features and EEG abnormalities showing an increased copy number within the region Xq28 containing a partial triplication

10.30 M.T. BONATI, F. VERDE, U. HLADNIK, L. CAMPANA, N. TICOZZI, L. MADERNA, C. COLOMBRITA, S. PAPA, P. BANFI AND V. SILANI
Occipital Horn syndrome in a 57 year-old man carrying a novel nonsense mutation in *ATP7A*: family report and literature review

10.45 A. VAN HAGEN, M. ELTING AND S. JANSEN
Mental retardation, (inner) ear malformations and dysmorphism; the expanding phenotype of MED12 related disorders

11.00-11.30 *Coffee Break*

11.30-12.30 FIFTH SESSION: Genetic testing in syndromology
Chair: GARAVELLI L. – PEETERS H.

11.30 A. SIFRIM, M.-P. HITZ, A. WILSDON, J. BRECKPOT, S.H. AL TURKI, B. THIENPONT, J. MCRAE, T.W. FITZGERALD, T.R SINGH, G. JAWAHAR SWAMINATHAN, E. PRIGMORE, D. RAJAN, H. ABDUL-KHALIQ, S. BANKA, U.M.M. BAUER, J. BENTHAM, F. BERGER, S. BHATTACHARYA, F. BU'LOCK, N. CANHAM, I.-G. COLGIU, C. COSGROVE, H.N COX, I. DAEHNERT, A. DALY, J. DANESH, A. FRYER, M. GEWILLIG, E. HOBSON, K. HOFF, T. HOMFRAY, THE INTERVAL STUDY, A.-K. KAHLERT, A. KETLEY, H.-H. KRAMER, K. LACHLAN, A.K. LAMPE, J.J. LOUW, A. KUMAR MANICKARA, D. MANASE, K.P. MCCARTHY, K. METCALFE, C. MOORE, R. NEWBURY-ECOB, S. OSMAN OMER, W.H. OUWEHAND, S.-M. PARK, M.J. PARKER, TH. PICKARDT, M. O POLLARD, L. ROBERT, D.J. ROBERTS, J. SAMBROOK, K. SETCHFIELD, B. STILLER, C. THORNBOROUGH, O. TOKA, H. WATKINS, D. WILLIAMS, M. WRIGHT, S. MITAL, P.E.F. DAUBENEY, B. KEAVNEY, J. GOODSHIP, THE UK10K CONSORTIUM, R. MAHDI ABU-SULAIMAN, S. KLAASSEN, C.F. WRIGHT, H.V. FIRTH, J.C. BARRETT, K. DEVRIENDT, D.R. FITZPATRICK, J.D. BROOK, THE DECIPHERING DEVELOPMENTAL DISORDERS STUDY AND M.E. HURLES
Distinct genetic architectures for syndromic and non-syndromic congenital heart defects identified by exome sequencing

11.45 Á. MARTÍN-RODRÍGUEZ, A. PRIETO-ABERASTURI, J. SÁNCHEZ AND A. GONZÁLEZ-MENESES
Usefulness of skin biopsy in the genetic diagnosis of mental retardation associated with skin pigmentary lesions

12.00 E. BIJLSMA, E. ATEN, D. FRANSEN VAN DE PUTTE, A. VAN HAERINGEN, N. DEN HOLLANDER, Y. HILHORST, S. KANT AND M. KOOPMANS, T. POTJER, M. HOFFER, C. RUIVENKAMP AND G. SANTEN
Unexpected diagnoses after whole exome sequencing

12.15 A. LUMAKA, V. RACE, H. PEETERS, P. LUKUSA AND K. DEVRIENDT
Clinical and molecular screening for the fragile X syndrome in specialized institutions in Kinshasa, DR Congo

AFTERNOON

14.30-15.45 SIXTH SESSION: Cytogenetics
Chair: CALLEWERT B. – KEYMOLEN K.

14.30 N. COSEMANS AND H. PEETERS
The clinical relevance of intragenic nrxn1 copy number variants

14.45 J. DE GEYTER, K. STEINDL, B. ONEDA AND A. RAUCH
New case confirms the role of CTNNB1 haploinsufficiency in intellectual disability

15.00 I. SCHANZE, S. BOPPUDI, S. BERLAND, M. GÉRARD, M. HOLDER-ESPINASSE, E. LACAZE, S. MORGAN, H. PEETERS, F. PETIT, D.T. PILZ, I. WIELAND, J.-B. RIVIÈRE, D. SCHANZE, L. FAIVRE AND M ZENKER
Haploinsufficiency of the *NFIB* gene in patients with mild intellectual disability

15.15 S.M. AZZARELLO-BURRI, P JOSET, J ANDRIEUX, F LOPES, E.E. PALMER, J.C. CZESCHIK, B. DEMEER, F. DUQUE, A. KÜCHLER, P. MACIEL, B. PLECKO, B. ONEDA AND A. RAUCH
Further Delineation of a novel 2q11.1q11.2 microduplication syndrome

15.30 R. NICOLESCU, J.-H. CABERG AND S. BULK
A family of Kagami-Ogata syndrome

15.45-16.15 *Coffee Break*

16.15-18.00 SEVENTH SESSION: Limb malformations
Chair: DEVRIENDT K.. – ZENKER A.

16.15 L. DARDOUR, J. BRECKPOT, P. MOENS, C. OOSTERLYNCK AND K. DEVRIENDT
FATCO syndrome: fibular hypoplasia, tibial campomelia and oligosyndactyly

16.30 P. THILOBO LUKUSA, L. DARDOUR, L. LEWI, P. MOERMAN AND K. DEVRIENDT
Confirmation of a distinct condition with severe limb deficiency, (mirror) polydactyly and variable organ malformations

16.45 A.T. MIDRO, J. BORYS, E. HUBERT, T. DAWIDZIUK, E. WASILEWSKA, J. SKOWRONSKI[4], E. TARASÓW, B. PANASIUK, E.M. OLECH AND A. JAMSHEER
TAR syndrome with orofacial clefting - 30 years follow–up

17.00 J KOHLHASE, G SCHMIDT AND W BOROZDIN
Baller-Gerold, RAPADILINO and Rothmund-Thomson syndromes: case reports and mutations detected in our patient cohort

17.15 D. HAYE, E. AMOUYAL, C. BAUMANN1, Y. CAPRI, S. ROSSIGNOL, M.-P. MOIZARD, A. VERLOES AND L. PERRIN
Hepatoblastoma in a female with Simpson Golabi Behmel Syndrome: should we monitor females with SGBS as we do for males ?

17.30 A. BOTTANI
On the potential importance for a dysmorphologist to (sometimes) attend a wine fair: a true story !