

# Friday, June 28, 2013

8:00 > 8:50 am Registration - Grand Hall  
Welcome coffee

## Conference room: GOYA AMPHITHEATER

8:50 > 9:00 am Welcome address  
*Stéphane Richard (Le Kremlin-Bicêtre, France)*

9:00 > 10:30 am **Session 1** - Chair: *Laura Schmidt, Bin Teh*

9:00 am **01** **FLCN-deficiency ablates plithotaxis, enhances cell-cell adhesion, and leads to cell-type specific defects in TGF-beta signaling**

*Damir Khabibullin, Doug Medvetz, Jae Hun Kim, Robert Steward Jr, Bomi Gweon, Erik Zhang, Andrey Parkhitko, Venkatesh Hariharan, Hayden Huang, Jane Yu, Jeffrey Fredberg, Elizabeth Henske*

9:30 am **02** **Exit from pluripotency is gated by intracellular redistribution of the bHLH transcription factor TFE3**

*Joerg Betschinger, Jennifer Nichols, Sabine Dietmann, Philip D. Corrin, Patrick J. Paddison, Austin Smith*

9:50 am **03** **Folliculin regulates lysosome function through mTORC1-dependent phosphorylation of transcription factor EB**

*Constance S. Petit, Agnès Roczniak-Ferguson, Shawn M. Ferguson*

10:10 am **04** **Constitutive AMPK activation upon loss of renal tumor suppressor folliculin leads to Warburg effect through HIF induction**

*Ming Yan, Marie-Claude Gingras, Elaine Dunlop, Anders Bondo Dydensborg, Rachael Preston, Zahra Jalali, Sylvie Sabourin, Yann Emmanuel Nouët, Brandon Faubert, Daina Avizonis, David Mark Davies, Taren Roughed, Dmitri Kharitidi, Russell Jones, Andrew Tee, Arnim Pause*

10:30 > 10:50 am Coffee break - Grand Hall

## Conference room: GOYA AMPHITHEATER

- 10:50 am > 12:10 pm **Session 2** - Chair: *Sophie Giraud, Eamonn Maher*
- 10:50 am **05** **Detection and functional characterization of new mutations in the FH gene**  
*Marine Guillaud-Bataille, Johnny Bombed, Abdel Slama, Michel Barrois, Sophie Deveaux, Patrick Benusiglio, INCa MCUL and HLRCC national group, Stéphane Richard, Brigitte Bressac-de Paillerets*
- 11:10 am **06** **Improved detection of FLCN mutations in patients with BHDS**  
*Jorge R. Toro, Bethany Friedman, Sherri Bale*
- 11:30 am **07** **BHD: a clinical and molecular study of 177 French patients**  
*Jérôme Lamoril, Nathalie Clément, Angélique Riffault, Agnès Bourillon, Caroline Kannengiesser, Bernard Grandchamp, Nadem Soufir*
- 11:50 am **08** **Germline BAP1 mutations predispose to renal cell carcinomas**  
*Tatiana Popova, Lucie Hebert, Sophie Gad, Stéphane Richard, Olivier Caron, Nadem Soufir, Brigitte Bressac-de-Paillerets, Dominique Stoppa-Lyonnet, Marc-Henri Stern*
- 12:10 > 12:40 pm **Invited lecture: New insights into the biology of clear cell renal carcinoma**  
*William Kaelin Jr (Harvard, USA)*
- 12:40 > 2:00 pm **Lunch and poster viewing - Grand Hall**

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2:00 > 3:10 pm **Session 3** - Chair: Marie-Françoise Avril, Masatoshi Kurihara

2:00 pm **09 Folliculin and lung: is it important?**

*Vera P. Krymskaya, Elena Goncharova, Melane L. Fehrenbach, Dmitry Goncharov, Seung-Beom Hong, Masaya Baba, W. Marston Linehan, Laura S. Schmidt*

2:30 pm **010 Folliculin regulates epithelial-mesenchymal transition in airway epithelium: implications for cyst formation in the BHD lung**

*Stephen Land, Claire Scott, Kendra Tosoni, Diane Cassidy, Stuart Booth, Anil Mehta*

2:50 pm **011 Folliculin gene-associated multiple cystic lung disease**

*Jean-François Cordier, Camille Taillé, Benoît Wallaert, Dominique Valeyre, the Groupe d'Etudes et de Recherche sur les Maladies "Orphelines" Pulmonaires (GERM"O"P)*

3:10 > 4:10 pm **Panel discussion: Clinical Guidelines for HLRCC**

*Fred Menko, Eamonn Maher, Laura Schmidt, Marston Linehan, Graham Lovitt, Stéphane Richard*

4:10 > 4:30 pm Coffee break - Grand Hall

## Conference room: GOYA AMPHITHEATER

4:30 > 5:50 pm **Session 4** - Chair: *Jean-François Cordier, Fred Menko*

4:30 pm **012 BHD versus non-BHD patients with spontaneous pneumothorax: chest CT findings**  
*Maartje J. Binnendijk, Paul C. Johannesma, Johan J.P. Gille, Arjan C. Houweling, Rinze Reinhard, Theo M. Starink, R. Jeroen A. van Moorselaar, Jan Hein van Waesberghe, Edward M. Leter, Quinten Waisfisz, Fred H. Menko, Piet E. Postmus*

4:50 pm **013 BHD patients with and without pneumothorax: chest CT findings**  
*Maartje J. Binnendijk, Paul C. Johannesma, Johan J.P. Gille, Arjan C. Houweling, Rinze Reinhard, Theo M. Starink, R. Jeroen A. van Moorselaar, Jan Hein van Waesberghe, Edward M. Leter, Quinten Waisfisz, Fred H. Menko, Piet E. Postmus*

5:10 pm **014 Thoracoscopic surgery for preventing recurrent pneumothorax in BHD syndrome: lower pleural covering technique (LPC)**  
*Masatoshi Kurihara, Hideyuki Kataoka, Kuniaki Seyama, Toshio Kumasaka*

5:30 pm **015 An update of a Birt-Hogg-Dubé syndrome (BHD) database: evaluation of 89 families referred for suspected BHD**  
*Fred H. Menko, Paul C. Johannesma, Johan J.P. Gille, Arjan C. Houweling, Edward M. Leter, Marianne A. Jonker, Yael Kon, Annelinde Terlouw, Theo M. Starink, Elisabeth H. Jaspars, Rinze Reinhard, Jan Hein van Waesberghe, R. Jeroen A. van Moorselaar, Piet E. Postmus, Cora Aalfs, Mirjam M. de Jong, Ernie MHF Bongers, Rogier A. Oldenburg, Theo A. van Os, Karin Y. van Spaendonck-Zwarts, Maurice A.M. van Steensel, Quinten Waisfisz*

5:50 > 6:20 pm **Invited lecture:**  
**Targeted therapies for advanced kidney cancer**  
*Bernard Escudier (Villejuif, France)*

# Friday, June 28, 2013

## Poster display: GRAND HALL

6:20 > 7:00 pm

### Poster viewing

**C1 Phenotypic and genotypic description of 10 BHD families from 2 distinct PREDIR regional Centers.**

*Caroline Abadie, Catherine Dugast, Brivael Gery, Carole Corsini, Sylvie Odent, Sophie Giraud, Stéphane Richard, Isabelle Coupier*

**C2 Radiofrequency ablation: an alternative treatment to surgery in Birt-Hogg-Dubé syndrome**

*Mohamed Karji Almarzoqi, Stéphane Richard, Dominique Joly, Arnaud Méjean, Olivier Hélénon, Jean-Michel Corréas*

**C3 Cutaneous and renal presentation of PTEN hamartoma tumor syndrome**

*Blandine Bel, Mathilde Funes de la Vega, Eric Mourey, Laurence Faivre, Michel Longy, Pierre Vabres*

**C4 Metastatic renal cell carcinoma in Birt-Hogg-Dubé patients**

*Patrick Benusiglio, Sophie Deveaux, Arnaud Méjean, Jean-Michel Corréas, Bernard Escudier, Stanislas Ropert, Ayhan Ulusakarya, Sophie Ferlicot, Sophie Giraud, Stéphane Richard*

**C5 A clinicopathologic study of 72 individuals in 33 Asian BHD families**

*Mitsuko Furuya, Reiko Tanaka, Masahiro Yao, Yoji Nagashima, Shunsuke Koga, Yukio Nakatani*

**C6 Folliculin is an evolutionary conserved regulator of AMPK function mediating metabolic stress survival through autophagy**

*Zahra Jalali, Yann Nouët, Elite Possik, Ming Yan, Marie-Claude Gingras, Laëtitia Chotard, Fanny Dupuy, Elea Prezel, Russell Jones, Richard Roy, Christian Rocheleau, David Hall, Arnim Pause*

**C7** Prevalence of Birt-Hogg-Dubé syndrome among patients with spontaneous pneumothorax: preliminary results

*Paul C. Johannesma, Maartje J. Binnendijk, Rinze Reinhard, Theo M. Starink, Yaël Kon, Annelinde Terlou, R. Jeroen A. van Moorselaar, Jan-Hein van Waesberghe, Arjan C. Houweling, Edward M. Leter, Quinten Waisfisiz, Johan J.P. Gille, Fred H. Menko, Pieter E. Postmus*

**C8** Exome sequencing and SNP array analysis of HLRCC syndromic uterine leiomyomas

*Kati Kämpjärvi, Miika Mehine, Pia Vahteristo, Lauri A. Aaltonen*

**C9** Interpretation of unclassified variants in the Fumarate Hydratase-(FH) gene by using immunohistochemistry for 2-succinyl cysteine (2SC)

*C. Marleen Kets, Christina A. Hulsbergen-van der Kaa, Maran J. Olderode-Berends, Fred H. Menko, Arjen R. Mensenkamp*

**C10** Malignant fibrous histiocytoma occurring in a previously undiagnosed Birt-Hogg-Dubé patient: a case-report

*Nicolas Poulalhon, Luc Thomas*

**C11** Exploring FLCN-HIF1 $\alpha$  cross-regulation: immunohistochemical and molecular studies

*Laura Maria Pradella, Claudia Ligorio, Giovanni Tallini, Marco Seri, Daniela Turchetti, Giuseppe Gasparre*

**C12** Pulmonary arterial hypertension in a patient with Birt-Hogg-Dubé syndrome and renal insufficiency

*Laurent Savale, Elise Artaud-Macari, Barbara Girerd, David Montani, Marc Humbert, Gérard Simonneau, Olivier Sitbon, Sophie Giraud, Stéphane Richard*

**C13** FLCN is involved in autophagy regulation

*Elaine A. Dunlop, Sara Seifan, Ewelina Rozycka, Ross Tomaino, Barry Coull, Ravi Nookala, Tijs Classens, James T. Murray, John Blenis, Maurice A.M. van Steensel, Simon Wilkinson, Andrew R. Tee*

## Friday, June 28, 2013

7:30 > 10:30 pm Conference dinner at the restaurant **Le First** with jazz concert: *Laurent Bataille (drums), Guillaume Naud (organ), Hugo Lippi (guitar)*.  
Business casual dress code

Meeting point at 7:15 pm in front of the Porte Jaujard (École du Louvre).

Walk from the École du Louvre to the restaurant Le First (15-minute walking distance).

Please remember to bring along your invitation card.

### Restaurant Le First

234, rue de Rivoli

or C/O the Westin Paris Hotel - 3, rue de Castiglione  
75001 PARIS

 1 Tuileries

# Saturday, June 29, 2013

8:00 > 9:00 am Registration - Grand Hall  
Welcome coffee

Conference room: MICHEL-ANGE AMPHITHEATER

9:00 > 10:30 am **Session 5** - Chair: Vera Krymskaya, Arnim Pause

9:00 am **016 Fumarate promotes ongoing pseudohypoxia through TBK1-mediated phosphorylation of RelA (p65)**

*Karthigayan Shanmugasundaram, EunHee Shim, Karen Block, Sunil Sudarshan*

9:30 am **017 HLRCC: insights from mouse models to man**

*Julie Adam, Ming Yang, Christina Bauerschmidt, Tomoyoshi Soga, Patrick J. Pollard*

9:50 am **018 Differential expression of CD44/DPYS in hereditary and sporadic Papillary Renal Cell Carcinoma type 2 (PRCC2) DPYS as the potential diagnostic biomarker**

*Victoria Perrier-Trudova, Aikseng Ooi, Sophie Gad, David Petillo, Vincent Molinié, Puay-Hoon Tan, Aye Aye Thike, Min-Han Tan, Betty Gardie, Stéphane Richard, Bin Tean-Teh,*

10:10 am **019 The role of mitochondrial DNA mutations in syndromic renal oncocytomas**

*Martin Lang, Cathy D. Vocke, Maria J. Merino, Laura S. Schmidt, W. Marston Linehan*

10:30 > 10:50 am Coffee break - Grand Hall



# Saturday, June 29, 2013

Conference room: MICHEL-ANGE AMPHITHEATER

- 10:50 am > 12:10 pm **Session 6** - Chair: *Elaine Dunlop, Damir Khabibullin*
- 10:50 am **020** **A FNIP1 binding protein identified by yeast two-hybrid screening links the FLCN/FNIP1 complex with membrane trafficking**  
*Masaya Baba, Hisashi Hasumi, Yukiko Hasumi, Ying Huang, Robert M. Hughes, Laura S. Schmidt, W. Marston Linehan*
- 11:10 am **021** **Folliculin and FNIP are evolutionary conserved regulators of AMPK function**  
*Arnim Pause, Yann Nouët, Zhara Jalali, Elite Possik, Ming Yan, Marie-Claude Gingras, Laëtitia Chotard, Fanny Dupuy, Elea Prezel, Russell Jones, Richard Roy, Christian Rocheleau, David Hall*
- 11:30 am **022** **FLCN is involved in autophagy regulation**  
*Elaine A. Dunlop, Sara Seifan, Ewelina Rozycka, Ross Tomaino, Barry Coull, Ravi Nookala, Tijs Classens, James T. Murray, John Blenis, Maurice A.M. van Steensel, Simon Wilkinson, Andrew R. Tee*
- 11:50 am **023** **Deletion of FLCN causes cardiac hypertrophy through AMPK-mTOR axis deregulation in mice**  
*Yukiko Hasumi, Masaya Baba, Hisashi Hasumi, Ying Huang, Antoine Smith, Hyounghbin Oh, Mara E. Klein, Robert M. Hughes, Kunio Nagashima, Laura S. Schmidt, Robert S. Adelstein, W. Marston Linehan*
- 12:10 > 12:40 pm Invited lecture: **Deep sequencing of HLRCC-related and sporadic type 2 Papillary Renal Cell Carcinoma: the search for tumorigenic and progression genes/pathways**  
*Aikseng Ooi, Bin Tean Teh (Grand Rapids, USA and Singapore, SG)*
- 12:40 > 2:00 pm Lunch and poster viewing - Grand Hall

## Conference room: MICHEL-ANGE AMPHITHEATER

2:00 > 3:20 pm **Session 7** - Chair: Sunil Sudarshan, Jorge Toro2:00 pm **024 Folliculin in organismal longevity and cellular stress resistance***Hakam Gharbi, Francesca Fabretti, Puneet Bharill, Markus Rinschen, Sibylle Brinkkötter, Peter Frommolt, Volker Burst, Bernhard Schermer, Thomas Benzing, Roman-Ulrich Müller*2:20 pm **025 Alterations in metabolic flux under FLCN deficiency***Hisashi Hasumi, Masaya Baba, Yukiko Hasumi, Ying Huang, Hyoungbin Oh, Robert M. Hughes, Mara E. Klein, Shoichi Takikita, Kunio Nagashima, Oksana Gavrilova, Samuel W. Cushman, Laura S. Schmidt, W. Marston Linehan*2:40 pm **026 Permanent genetic correction of the phenotype of BHD cells using episomally maintained DNA vectors***Suet Ping Wong, Richard P. Harbottle*3:00 pm **027 Efficacy of AZD8055, a dual mTORC1/mTORC2 kinase inhibitor, in the Nihon rat tumor model of BHD***Laura S. Schmidt, Lilia V. Ileva, Joseph D. Kalen, Diana C. Haines, Daniel L. Logsdon, Craig L. Driver, Amy M. James, Yukiko Hasumi, Masaya Baba, Sylvie Guichard, Ramaprasad Srinivasan, W. Marston Linehan*3:20 pm Late breaking communication  
**Birt-Hogg-Dubé Syndrome is a ciliopathy***Monique N. H. Luijten<sup>1,+</sup>, Sander G. Basten<sup>2,3,+</sup>, Tijs Claessens<sup>1,5,+</sup>, Marigje Vernooij<sup>1</sup>, Claire L. Scott<sup>4</sup>, Renske Janssen<sup>1</sup>, Jennifer A. Easton<sup>1</sup>, Miriam A. F. Kamps<sup>1</sup>, Maaïke Vreeburg<sup>6</sup>, Jos L. V. Broers<sup>7</sup>, Michel van Geel<sup>1</sup>, Fred H. Menko<sup>8</sup>, Richard P. Harbottle<sup>9</sup>, Ravi K. Nookala<sup>10</sup>, Andrew R. Tee<sup>5</sup>, Stephen C. Land<sup>4</sup>, Rachel Giles<sup>2</sup>, Barry J. Coull<sup>1,\*</sup>, Maurice A. M. van SteENSEL<sup>1,6</sup>*

3:30 &gt; 3:40 pm Closure of the Symposium

Organized by / organisée par *Sophie Deveaux, PhD*  
 and / et *Lindsay Middleton, R.N.*  
 Genetic counsellors / *Conseillères en Génétique*  
 Translation / traduction : *Sophie Deveaux, David Lipson*

## Saturday, June 29, 2013 / Samedi 29 juin 2013

8:00 > 9:00 am Registration / Inscription

9:00 > 9:20 am Welcome address / Message de bienvenue  
*Sophie Deveaux, Stéphane Richard (Le Kremlin-Bicêtre, France)*

9:20 > 10:30 am **Session 1** - Chair: *Caroline Abadie, Eamonn Maher*

**Role of patient advocacy groups and the benefits of collaboration /**

**Rôle des associations de patients et intérêt des collaborations**

*Ilene Sussman (Boston, USA)*

**BHD: the pulmonary perspective in relation to pneumothorax /**

**Atteinte pulmonaire du syndrome de Birt-Hogg-Dubé**

*Piet E Postmus (Amsterdam, NL)*

10:30 > 10:50 am Coffee break / Pause café

10:50 am > 12:40 pm **Session 2** - Chair: *Patrick Benusiglio, Ilene Sussman*

**A review of FLCN and FH germline mutations /**

**Le point sur les mutations germinales FLCN et FH**

*Jorge R. Toro (Rockville, USA)*

**Renal cell cancer in BHD and HLRCC: risk counseling and surveillance /**

**Cancer rénal, BHD et léiomyomatose : risque et surveillance**

*Fred Menko (Amsterdam, NL)*

Interactive discussion / Discussion interactive

12:40 > 14:00 pm Lunch and poster viewing / Déjeuner et visite des posters