

## Thuy-Linh Le

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## EDUCATION

- 2020            **Ph.D.** in Genetics, Université de Paris, France  
Thesis: Genetics of Hirschsprung Disease  
Advisor: Prof. Stanislas Lyonnet
- 2016            **M.S.** in Genetics, Paris Descartes University, France  
Thesis: Oligogenic model of Isolated form of Hirschsprung Disease  
Advisor: Prof. Stanislas Lyonnet
- 2014            **M.B.B.S.**, Hanoi Medical University, Vietnam  
Thesis: Laparoscopic cholecystectomy as a treatment of Acute cholecystitis  
Advisor: Prof. Bao-Long Tran

## EXPERIENCES

- 2020 - present    **Postdoctoral Researcher**, Institut Pasteur, Paris, France  
Human Genetics and Cognitive Function Laboratory  
Team leader: Prof. Thomas Bourgeron
- 2016 - 2020      **PhD Candidate**, Institut *Imagine*, INSERM UMR1163, Paris, France  
Embryology and Genetics of Malformations Laboratory  
Team leader: Prof. Jeanne Amiel

## SKILLS

- Wet lab            PCR, RT-qPCR, DNA sequencing, *In vitro* RNA synthesis,  
Site-directed mutagenesis, Cell culture, Plasmid transfection,  
Western Blot, Immunohistochemistry, IncuCyte® live-cell imaging
- Zebrafish genome editing by morpholino and CRISPR/Cas9  
                      Zebrafish and mouse mutant phenotype analysis –  
                      Dissection, Histopathology and Confocal Microscopy/Spinning Disk
- Dry lab            Exome analysis – Variant prioritisation and Segregation study  
*In silico* mutagenesis and protein crystal structure analysis (Chimera)  
Image analysis (ImageJ, Imaris), Data analysis and visualisation (R)
- Programming    C, SQL, Python
- Languages        Vietnamese (native), French (DALF C1), English (IELTS 7.5)

## FUNDINGS

2020	Fellowship of Excellence for Young Investigators, ESHG Conference
2019 - 2020	PhD Fellowship, Fondation pour la Recherche Médicale
2016 - 2019	PhD Fellowship, French Embassy in Vietnam
2015 - 2016	MS scholarship, French Embassy in Vietnam
2014 - 2015	MS scholarship, Agence Universitaire de la Francophonie

## INVITED TALKS

"Mapping and Identifying Genetic Susceptibility to Hirschsprung Disease."

Graduate course: Human Genetics and Pathologies, Université de Paris, Jan 27, 2021.

"New genes in syndromic Hirschsprung Disease involve the NRG1-ERBB and SHH pathways."

Imagine Seminar, Institut *Imagine*, Oct 26, 2020.

## PRESENTATIONS

"Biallelic mutations of *SMO* in humans cause a broad spectrum of developmental anomalies due to abnormal Hedgehog signalling."

ESHG Conference 2020, June 6-9, 2020, oral presentation.

"Biallelic mutations in *ERBB3* cause variable defects in multiple neural crest derivatives in humans."

YR2I Congress, Paris, France, May 23, 2019, oral presentation.

GRS-GRC Neural Crest and Cranial Placodes, Lucca, Italy, April 14-19, 2019, poster.

ENS Development Meeting, Boston, USA, April 8-11, 2018, poster.

"New generation sequencing allows identification of rare variants in the oligogenic model of Hirschsprung disease."

Assises de Génétique Humaine et Médicale 9, Nantes, France, Jan 24-26, 2018, poster.

Assises de Génétique Humaine et Médicale 8, Lyon, France, Feb 3-5, 2016, poster.

## PUBLICATIONS

- [1] **Thuy-Linh Le** et al. Dysregulation of the NRG1-ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. *The Journal of Clinical Investigation*, January 2021.
- [2] **Thuy-Linh Le** et al. Bi-allelic Variations of *SMO* in Humans Cause a Broad Spectrum of Developmental Anomalies Due to Abnormal Hedgehog Signaling. *American Journal of Human Genetics*, 106(6):779–792, June 2020.
- [3] Hongsheng Gui et al. Whole exome sequencing coupled with unbiased functional analysis reveals new Hirschsprung disease genes. *Genome Biology*, 18(1):48, March 2017.
- [4] Clara Sze-Man Tang et al. Trans-ethnic meta-analysis of genome-wide association studies for Hirschsprung disease. *Human Molecular Genetics*, 25(23):5265–5275, December 2016.
- [5] Thi Lua Nguyen et al. Survey-based cancer mortality in the Lao PDR, 2007-08. *Asian Pacific journal of cancer prevention*, 12(10):2495–2498, 2011.

## REFEREES

Prof. Stanislas Lyonnet  
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Professor, Université de Paris  
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Research Director, Institut *Imagine*  
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Prof. Jeanne Amiel  
Team leader, Institut *Imagine*  
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Dr. Sophie Thomas  
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