POSTDOCTORAL RESEARCHER (m/f)

The job offer is located in the inserm 1163 laboratory at Imagine institute, whose aim is to characterize and treat genetic diseases and offers facilities and platforms to carry out your research

A funding for 2 year post-doctoral position is available from **September 2024** in the research group of Pr Valérie Cormier-Daire, "Molecular and physiopathological bases of osteochondrodysplasia" at the Imagine Institute (www.institutimagine.org). The research team is especially interested in studying the molecular, genetic, pathophysiological mechanisms and treatments of the different types of osteogenesis imperfecta (OI). These disabling diseases are characterized by bone fragility, fractures, short stature, and skeletal deformities. Our aims are to further understand the pathophysiology of these rare disorders and develop novel perspectives in their treatments. Our project has two main goals

1) to study the pathophysiology of OI type V related to IFTM5 variant by performing Omics approaches in human cells
2) to test the effect of neutralizing anti-TGF beta treatment in OI mouse model, mimicking OI type 1. We hypothesize that low-dose treatment for a long time may be safe for children and may improve growth defect and bone fragility.. The postdoc researcher will study the effects of the treatment in murine osteogenesis imperfecta at early ages and will complete by investigating the roles of collagen receptors in the short stature phenotype.

Required skills*:* Expertise in molecular and cellular biology. Expertise in animal facilities and mouse experiments. The candidate must be able to work independently, be dynamic, communicate and work effectively with the team and internal and external collaborators. The candidate must be rigorous, organized and work methodically and critically. He/she must be interested in technological development and translational research. He/she must also be able to supervise and train students working with him/her on the project.

Type of contract and annual salary: 12-month fixed-term contract starting **September 15, 2024** (renewable up to 2 years) and salary depending on experience.

Applications in English including a cover letter, a detailed CV with publication list and references should be sent to Pr V Cormier-Daire (valerie.cormier-daire@inserm.fr)

Selected publications of the team:

1. Dubail J, Huber C, Chantepie S, Sonntag S, Tüysüz B, Mihci E, Gordon CT, Steichen-Gersdorf E, Amiel J, Nur B, Stolte-Dijkstra I, van Eerde AM, van Gassen KL, Breugem CC, Stegmann A, Lekszas C, Maroofian R, Karimiani EG, Bruneel A, Seta N, Munnich A, Papy-Garcia D, De La Dure-Molla M, **Cormier-Daire V.** SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects.
Nat Commun. 2018, 9:3087

2.Doyard M, Bacrot S, Huber C, Di Rocco M, Goldenberg A, Aglan MS, Brunelle P, Temtamy S, Michot C, Otaify GA, Haudry C, Castanet M, Leroux J, Bonnefont JP, Munnich A, Baujat G, Lapunzina P, Monnot S, Ruiz-Perez VL, **Cormier-Daire** V. *FAM46A* mutations are responsible for autosomal recessive osteogenesis imperfecta.
J Med Genet. 2018, 55: 278-284

3. Dubail J, Brunelle P, Baujat G, Huber C, Doyard M, Michot C, Chavassieux P, Khairouni A, Topouchian V, Monnot S, Koumakis E, **Cormier-Daire** V Homozygous loss-of-function mutations in CCDC134 are responsible for a severe form of osteogenesis imperfecta. ***J Bone Miner Res. 2020, 35 : 1470-1480***

4. Marzin P, Thierry B, Dancasius A, Cavau A, Michot C, Rondeau S, Baujat G, Phan G, Bonnière M, Le Bourgeois M, Khraiche D, Pejin Z, Bonnet D, Delacourt C, **Cormier-Daire V**. Geleophysic and acromicric dysplasias: natural history, genotype-phenotype correlations, and management guidelines from 38 cases.
Genet Med. 2021, 23 : 331-3

5. Guasto A, Dubail J, Aguilera-Albesa S, Paganini C, Vanhulle C, Haouari W, Gorría-Redondo N, Aznal-Sainz E, Boddaert N, Planas-Serra L, Schlüter A, Verdura E, Bruneel A, Rossi A, Huber C, Pujol A, Cormier-Daire V. Biallelic variants in SLC35B2 cause a novel chondrodysplasia with hypomyelinating leukodystrophy.
Brain. 2022, 145 : 3711-3722