# Japanese post-doctoral fellowship at the Institut Pasteur: first call (2019)

Research project 4

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| **Host laboratory** | Stability of Nuclear and Mitochondrial DNA Team, Stem Cells and Development Unit |
| **Contact** | boursepdjapon@pasteur.fr |
| **Title** | Underlying mechanisms and therapeutic perspectives for progeroid Cockayne syndrome |
| **Abstract** | Cockayne syndrome (CS) is a genetic disease characterized by progeroid defects, neurodegeneration, and frequently UV/[sun]-hypersensitivity. We identified a defective pathway in CS patient cells that leads to alterations in mitochondrial function. We rescued the defective pathway with a molecule, MnTBAP that obtained Orphan Drug Designation for CS. Importantly, this rare disease displays a large diversity of clinical symptoms, the mechanistic and genetic reasons of which are not understood. CS is well studied in Europe and Japan, where particularly interesting cases have been identified. Using selected patient-derived cells, iPSCs, organoids, and ongoing isogenic models, we plan to assess the link between mitochondrial dysfunction and the severity of CS, in the context of different genetic backgrounds, and with a therapeutic perspective (personalized medicine). The post-doc's work will be fundamental for establishing/reinforcing tighter links between Institut Pasteur and Japanese teams on CS, from an original perspective that has also implications for regular ageing. |