3D Human Axial Development In Vitro: using novel human in vitro somitogenesis models to study birth defects with patient-relevant iPS cell lines

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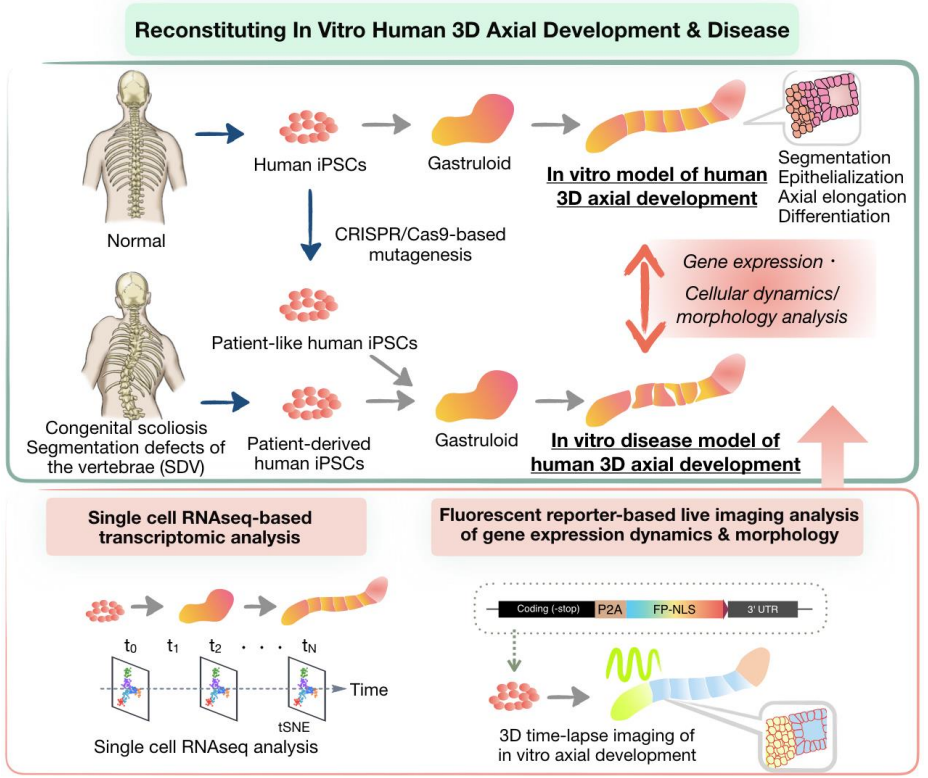
Congenital scoliosis and segmentation defects of the vertebrae (SDV) are medical conditions characterized by abnormal formation and patterning of the spine and axial skeleton, thought to be caused by defects during somitogenesis and axial development of the early embryo. Although it is highly warranted to study human somitogenesis for proper understanding of normal as well as abnormal human development, analyzing human embryos is difficult due to ethical and technical restrictions. Here we propose the establishment of an induced pluripotent stem cell (iPSC)-based in vitro model of human 3D axial development & disease, aiming to characterize the functional and molecular features of human somitogenesis and to study the pathogenesis of congenital defects of the spine and axial skeleton. To this end we will utilize our newly established in vitro 3D axial development model in combination with patient-derived and patient-like iPSCs containing pathogenic mutations. We will analyze and compare in vitro human 3D axial development and somitogenesis under normal and disease conditions by utilizing single cell RNAseq-based transcriptomic analysis and fluorescent live imaging-based assessment of gene expression dynamics and morphological changes. Successful implementation of our joint research project will increase our still limited understanding of human development and disease.



■URL: https://ashbi.kyoto-u.ac.jp/ja/groups/alev/