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Upstream of precise disease models for better downstream decision making

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Inborn errors of metabolism are a common cause of inherited diseases. Diseases of carbohydrate metabolism pathway include lysosomal storage diseases (LSDs), which are a significant subgroup with approximately 70 LSDs. Grouped according to their defective lysosomal proteins and pathways they are usually characterized by intralysosomal accumulation of substrate. Accumulation may occur at different levels in diverse types of cells, some of which are of difficult access. Patient, molecular, cell and tissue heterogeneity hinders the development of further therapeutic approaches.

We are currently establishing human Induced Pluripotent Stem Cells (iPSCs) from fibroblasts of LSDs patients and controls. The use of disease-specific cell models, mimicking the cell-target of the specific disease, may help to appropriately study the pathogenesis as well as the therapeutics. Integrating new techniques in the work pipeline for the establishment of models may lead to more accurate models while ensuring the safeguard of the patient's background.

Advanced technologies like microarray and NGS profiling add to the traditional techniques such as Immunofluorescence, directed sequencing and conventional cytogenetics. As in the diagnosis process, we may better understand the prognosis, and contribute to cost avoidance, by combining genomic and protein profiling checkpoints in the cell-model establishment pipeline.

The investment in the upstream checkpoints might prove to be helpful in ensuring the integrity of the cell models.