S07-3 Identification of novel transporter diseases of connective tissue and clarification of their molecular mechanism by combined approach of mouse and human genetics

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Mouse and human are valuable research resource for disease study. These “model animals” are good tools for genetic and genomic approaches for skeletal diseases. Because both have pros and cons, we are using a combined approach of mouse and human genetics. Recently, we have identified disease genes of two skeletal diseases; both are caused by the mutations in genes encoding transporters of previously unknown biological function. One is a nucleotide–sugar transporter on ER and another a Zn transporter on Golgi. Our discovery would open a new window for study of skeletal diseases.