## Logical Axioms for Inheritance in the DO

Import ontologies:

GENO [at EBI OLS] for inheritance

SO -- for structural variants (e.g. genetic disease, monogenic disease)

The EQ statements have already been added to the DO.

## **EQ** statements

genetic disease

monogenic

autosomal genetic disease autosomal dominant disease autosomal recessive disease disease and ('disease has basis in' some structural\_variant)

disease and 'disease has basis in' some gene

disease and ('has material basis in' some 'autosomal inheritance')

disease and ('has material basis in' some 'autosomal dominant inheritance') disease and ('has material basis in' some 'autosomal recessive inheritance')

X-linked monogenic disease

disease and ('has material basis in' some 'X-linked inheritance')

X-linked recessive disease disease and ('has material basis in' some 'X-linked recessive inheritance')
X-linked dominant disease disease and ('has material basis in' some 'X-linked dominant inheritance')

The SubClass Of Statements are added to DO terms, when the asserted parent is outside of The genetic disease branch:

genetic disease

Chromosomal disease

▶ ☐ Alport syndrome

Opitz-GBBB syndrome

X-linked monogenic disease

Y-linked monogenic disease

autosomal genetic disease

The SubClass of Statements include:

The EQ statements have already been added to the DO.

## **SubClass Of statements**

genetic disease 'disease has basis in' some structural\_variant

monogenic 'disease has basis in' some gene

autosomal genetic disease 'has material basis in' some 'autosomal inheritance'

autosomal dominant disease 'has material basis in' some 'autosomal dominant inheritance' autosomal recessive disease 'has material basis in' some 'autosomal recessive inheritance'

X-linked monogenic disease 'has material basis in' some 'X-linked inheritance'

X-linked recessive disease 'has material basis in' some 'X-linked recessive inheritance'
X-linked dominant disease 'has material basis in' some 'X-linked dominant inheritance'