DISEASE:
Situs ambiguus

NAME:	Situs ambiguus
DESCRIPTION:	A rare, genetic, developmental defect during embryogenesis characterized by a partial mirror-image transposition of intra-thoracic and/or intra-abdominal organs across the left-right axis of the body. Intra-organ variations and other malformations, such as ciliary motricity anomalies (e.g. Kartagener syndrome), biliary atresia and cardiac defects, are frequently associated. Left (polysplenia syndrome) or right (asplenia syndrome) isomerism are usually observed.
ORPHACODE:	157769
SYNONYMS:	Incomplete situs inversus Partial situs inversus Situs ambiguous
XREF(S):	Orphanet ICD-10 MedDRA

ANALYTE(S):	PKD1L1 ZIC3 ACVR2B LEFTY2 NODAL CFAP53 MMP21 NODAL DNAH9 CFC1
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- matrix metallopeptidase 21
- nodal growth differentiation factor
- polycystin 1 like 1, transient receptor potential channel interacting
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