**Animal Bioresource in Japan**

# Characterization of the Skeletal Fusion with Sterility (sks) Mouse Showing Axial Skeleton Abnormalities Caused by Defects of Embryonic Skeletal Development

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**Abstarc :**

The development of the axial skeleton is a complex process, consisting of segmentation and differentiation of somites and ossification of the vertebrae. The autosomal recessive skeletal fusion with sterility (*sks*) mutation of the mouse causes skeletal malformations due to fusion of the vertebrae and ribs, but the underlying defects of vertebral formation during embryonic development have not yet been elucidated. For the present study, we examined the skeletal phenotypes of *sks*/*sks* mice during embryonic development and the chromosomal localization of the *sks* locus. Multiple defects of the axial skeleton, including fusion of vertebrae and fusion and bifurcation of ribs, were observed in adult and neonatal *sks*/*sks* mice. In addition, we also found polydactyly and delayed skull ossification in the *sks*/*sks* mice. Morphological defects, including disorganized vertebral arches and fusions and bifurcations of the axial skeletal elements, were observed during embryonic development at embryonic day 12.5 (E12.5) and E14.5. However, no morphological abnormality was observed at E11.5, indicating that defects of the axial skeleton are caused by malformation of the cartilaginous vertebra and ribs at an early developmental stage after formation and segmentation of the somites. By linkage analysis, the *sks* locus was mapped to an 8-Mb region of chromosome 4 between *D4Mit331* and *D4Mit199*. Since no gene has already been identified as a cause of malformation of the vertebra and ribs in this region, the gene responsible for *sks* is suggested to be a novel gene essential for the cartilaginous vertebra and ribs.

**Key Word :**
mapping, mutant mouse, skeletal defect

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