Oculoskeletal Dysplasia 2 or Dwarfism- Retinal Dysplasia 2, (OSD2)

In brief
Oculoskeletal Dysplasia is a disorder causing both vision and skeletal abnormalities (short-limbed dwarfism) in Labrador Retrievers (OSD1) and in Samoyeds (OSD2). Affected dogs have short limbs, retinal hypoplasia and other eye defects. The disease is caused by mutations in the COL9A2 or COL9A3 genes and inherited in an autosomal recessive manner.

Clinical overview
Oculoskeletal Dysplasia is a disorder causing alterations to vision and limbs. Affected puppy’s limbs, especially forelimbs are abnormally short and turned into an abnormal position. This can lead to developmental elbow disorder and osteoarthritis. Also, the skull shape of the affected dog is abnormal. The affected dog’s develop various eye abnormalities, such as vitreous dysplasia, retinal detachment and cataracts. Affected puppies can be distinguished from their littermates at the age of 4 to 6 weeks by their appearance. Eye abnormalities are frequently present at the age of 6 weeks. The above described symptoms develop to affected puppy that has two copies of the mutated gene, one from each parent. In addition, some carriers heterozygous for the causative mutation may develop eye defects but no skeletal abnormalities.

References
Online database
Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/), Faculty of Veterinary Science, University of Sydney, April 2013, OMIA001522-9615 and OMIA001523-9615.

Scientific articles