Bruck Syndrome 1

**Alternative Names**
- BRKS1
- Osteogenesis Imperfecta-Congenital Joint Contractures Syndrome
- Kuskokwim Disease
- Arthrogryposis-Like Disorder

**Record Category**
Disease phenotype

**WHO-ICD**
Diseases of the musculoskeletal system and connective tissue > Other specified acquired deformities of limbs

**Incidence per 100,000 Live Births**
<1

**OMIM Number**
259450

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
17q21.2

**Description**
Bruck syndrome is a rare autosomal recessive condition. It is characterized by a combination of congenital joint contractures and osteogenesis imperfecta. The onset of fractures becomes evident in infancy or early childhood. Few cases (<40) have been reported in the literature. The symptoms of Bruck syndrome include osteoporosis and bone fragility, progressive joint contractures which may be seen with pterygia, wormian bones, scoliosis due to vertebral deformities and postnatal short stature. Mental development of the patients is intact.

**Molecular Genetics**
Two distinct genetic loci have been linked to Bruck Syndrome. One of these, located on the long arm of chromosome 17, maps to the FKBP10 gene. The other locus maps to the PLOD2 gene and is referred to as giving rise to Bruck Syndrome 2. The FKBP10 gene contains 10 exons and the encoded protein contains 582 amino acids. FKBP10 is thought to function as a collagen chaperone and to assist in collagen folding.

**Epidemiology in the Arab World**

**Saudi Arabia**
Shaheen et al. (2011) described two Saudi Arabian families with Bruck Syndrome. Parents in both families were consanguineous. The two affected sisters in family 1 had skeletal deformities, short stature and delayed development. Mental development was normal in both. Examination of heart, lung, abdomen and central nervous system was unremarkable. Pamidronate was given to the second sister at the age of 5-years and she showed a significant improvement, as she was described as fracture-free for 4 years. The affected children from the second family were cousins of patients in family 1. The index case in family 2 was a 17-year-old boy. He developed the first fracture during difficult labor. He had frequent fractures (3 per year), never walked and developed scoliosis. His brother developed fractures at age of 3 years and improved when pamidronate was administered. Shaheen et al., (2011) identified p.Gln249ThrfsX12 mutation in family 1 and p.Gly278ArgfsX95 mutation in family 2, in the FKBP10 gene. The authors affirm that mutations in FKBP10 gene cause Bruck syndrome and isolated osteogenesis imperfect.

**References**

**Related CTGA Records**
FK506-Binding Protein 10

**External Links**
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2771

**Contributors**
Ameera Balobaid: 05.06.2016