Bohring-Opitz Syndrome

Main clinical craniofacial features
(Different in expression and seriousness)

- Brain abnormalities as Agenesis Corpus Callosum
- Microcephaly (small head)
- Trigonocephaly (triangular shaped forehead)
- Broad prominent forehead
- Low posterior hairline
- Hypertrichosis (abnormal amount of hair growth)
- Low-set, posteriorly rotated ears
- Cleft lip/palate
- High narrow palate with prominent ridges
- Micro- or retrognathia (small and/or recessed jaw)
- Glabellar nevus simplex (birthmark on forehead)
- Exophthalmos (bulging eyes) with high myopia
- Up slanting palpebral fissures
- Retinal/optic nerve abnormalities
- Ocular Hypertelorism (large distance between eyes)
- Depressed and wide nasal bridge
- Anteverted nares (nasal openings are tipped upwards)
Bohring-Opitz Syndrome
Main clinical features
(Different in expression and seriousness)

- IUGR (intrauterine growth retardation)
- Severe intellectual and motor disabilities
- Failure to thrive
- Hypotonia (low muscle tone)
- Hypertrichosis
- BOS posture of the upper limbs (defined as having three out of four features: exorotation and/or adduction of the shoulders; flexion at the elbows; flexion at the wrists; and ulnar deviation of the wrists and/or fingers at the metacarpophalangeal (MCP) joints)
- Fixed contractures
- Congenital talipes equinovarus (deformity of the feet)
- Apneas
- Silent respiratory
- Recurrent infections
- Cardiac abnormalities
- Wilms tumor
- Renal abnormalities
- Severe feedings difficulties
- Intestinal malrotation
- Deep palm creases
- Absence of distal interphalangeal creases from fingers
- Thoracic kyphosis
- Hip dysplasia
- Spina bifida occulta
- Inguinal hernia
- Genital abnormalities
- Seizures
- Wilms tumor
- Renal abnormalities
- Severe feedings difficulties
- Intestinal malrotation