Bohring-Opitz syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Bohring-Opitz syndrome. ORPHA:97297

Bohring-Opitz syndrome is characterised by intrauterine growth retardation (IUGR), failure to thrive, facial dysmorphism (prominent metopic suture and forehead nevus flammeus, a low frontal and temporal hairline with hirsutism, puffy cheeks, upslanting palpebral fissures, exophthalmos, hypertelorism, cleft lip and palate, retrognathia and low set ears), flexion deformities of the elbows and wrists, camptodactyly, ulnar deviation of the fingers, foot anomalies and severe developmental delay. Less than 20 patients have been described so far. Although the large majority of reported cases occurred sporadically, autosomal recessive inheritance has also been reported.