

Leri-Weill Dyschondrosteosis: Diagnosis and Management

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Leri-Weill dyschondrosteosis (LWD), is a genetic disorder, inherited as autosomal dominant pattern. Is characterized by skeletal dysplasia with disproportionate short stature. There is a mesomelic shortening of the forearm and lower leg. SHOX mutations are responsible for 70% of cases, with the prevalence of 1/1000. The diagnosis is based on clinical, radiologic and molecular analysis for confirmation. The patients showed disproportionate short stature, short arms and lower legs. The skeletal x-ray confirmed the clinical observation. Madelung deformity of the arm is found in 74% of children. The wrist grows abnormally and part of radius stops growing. Is bilateral in 50 - 66% of cases. This deformity can appear also in Turner syndrome. One of the parents can showed same phenotype. Growth hormone deficiency can be associated with LWD. The differential diagnosis is with other SHOX-related haploinsufficiency disorders. If the mother is affected, the deletion of the SHOX gene is inherited in the pseudoautosomal region of X chromosome. The daughters of the index case will inherited the X chromosome of the father and will be affected. The boys will inherited the Y chromosome of the father. Prenatal diagnosis and genetic counseling are available for this syndrome. We can use growth hormone treatment to improve final adult height. If the patient has Madelung deformity, wrist ergonomic devices is indicated. Surgical intervention is required in some situations.

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