

Dysmorphic Syndromes with Skeletal Anomalies

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The dysmorphic syndromes consist of a group of polymalformative diseases, in which the association of several congenital deformities involving different organs and systems is repeated with such a frequency as to exclude the occasional moment and especially when it is demonstrated that all the anomalies are transmitted with hereditary or familiar character.

In some of these malformative diseases, the skeleton is affected in a constant or frequent and, often, characteristic manner. The subdivision of the different syndromes with skeletal involvement into various groups following a criterion that favors the most constant and prominent symptom allows to elaborate a classification, which has no claim to scientific rigor, but only aims to allow easier memorization of the syndromes themselves for the researcher.

We will divide them into

- · Syndromes with lethal or sublethal evolution
- Syndromes with craniofacial anomalies
- Syndromes with anomalies of the skull and extremities
- · Syndromes with anomalies of the face and extremities
- Syndromes with anomalies of the oral cavity and extremities
- Syndromes with prevalent involvement of the trunk skeleton

- Syndromes with prevalent involvement of the skeleton of the limbs
- Syndromes with joint hypermobility
- Syndromes with joint stiffness
- · Syndromes with bone overgrowth
- · Syndromes with dolichostenomelia
- · Syndromes with congenital gigantism
- Syndromes with prenatal dwarfism and skeletal anomalies
- Syndromes with cerebral and skeletal anomalies
- · Syndromes with ocular and skeletal anomalies
- · Syndromes with cardiac and skeletal anomalies
- Syndromes with pulmonary and skeletal anomalies
- · Syndromes with genital and skeletal anomalies
- · Syndromes with renal and skeletal anomalies
- · Syndromes with skin and skeletal anomalies
- · Syndromes with pterygium and skeletal anomalies
- Syndrome with progeroid status and skeletal anomalies
- Syndromes that associate anemia or thrombocytopenia and skeletal defects
- Skeletal syndromes resulting from metabolic abnormalities
- Syndromes involving nerve transmission to the muscles

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