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NUEVAS TERAPIAS ENDOCRINOLOGÍA PEDIÁTRICA

Vosoritide in the treatment of achondroplasia

Vosoritida en el tratamiento de la acondroplasia

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Achondroplasia is the most common type of skeletal dysplasia, affecting 1 in 20,000 individuals worldwide. It is caused by a recurrent, activating pathogenic variant in *FGFR3*, the gene that encodes the fibroblast growth factor receptor type 3 (FGFR3), a key component of the cartilaginous growth plate. Since the role of the FGFR3 signalling pathway is to inhibit endochondral bone growth, the skeletal effects of achondroplasia are widespread, causing disproportionate short stature, bone deformities, spinal anomalies and multiple associated medical problems. The most severe manifestation is stenosis of the foramen magnum, which if not detected leads to a fiftyfold increase in sudden infant death.

Vosoritide is a synthetic analogue of C-natriuretic peptide (CNP), the ligand that triggers another growth plate-active signalling pathway, the natriuretic peptide receptor type B (NPRB). Activation of NPRB counteracts the effects of the FGFR3 signalling pathway in achondroplasia.

This presentation will cover the multisystem and lifelong nature of achondroplasia, and the best-practice approach to management. It will highlight the outcomes of the extensive clinical trial programme and the transformative impact being realised through treatment with vosoritide today. In addition, there will be a discussion of the future potential uses of this treatment in related conditions.