Open Peer Review on Qeios

Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Short</u> <u>stature-deafness-neutrophil dysfunction-dysmorphism syndrome</u>. ORPHA:2866

Short stature-deafness-neutrophil dysfunction-dysmorphism syndrome is characterised by short stature, sensorineural deafness, mutism, facial dysmorphism and abnormal neutrophil chemotaxis (leading to recurrent infections).