

Sphenoid asymmetry associated to other skeletal anomalies in a clear cut case of *PTCH1* mutated Gorlin-Goltz syndrome- a novel finding?

C. Ruini¹, L. Pastorino², M. Malagoli³, W. Bruno², P. Ghiorzo², G. Bianchi-Scarrà², P. Loschi⁴, G. Pellacani¹, A. Tomasi³, F. Farnetani¹, V.D. Mande¹, G. Ponti³

¹Dip. Dermatologia, Università di Modena e Reggio Emilia; ²Genetica dei Tumori Ereditari Rari, DiMI Università degli Studi di Genova, IRCCS AOU San Martino-IST; ³Dip. di Medicina Diagnostica, Clinica e di Sanità Pubblica, Università di Modena e Reggio Emilia; ⁴Dip. Chirurgia Plastica e Ricostruttiva, Università di Modena e Reggio Emilia.

Gorlin-Goltz syndrome is an autosomal dominantly inherited disorder linked to *PTCH1* mutation, recognized by a collection of clinical and radiologic signs (macrocephaly, frontal bossing, multiple intracranial calcifications including falx cerebri and atlanto-occipital ligament). We describe here a the case of a family with clear cut criteria for Gorlin-Goltz syndrome presenting the association of cranio-facial and skeletal anomalies together with a peculiar sphenoid variant.

Two patients, father and son, were examined because of multiple basal cell carcinomas and keratocystic odontogenic tumours. Other suggestive findings were multiple positive family history, typical skeletal anomalies and a novel *PTCH1* germline mutation (c.1041delAA). Craniofacial and other skeletal anomalies displayed at 3D and helical CT scan were: macrocephaly, skull base asymmetry (positional plagiocephaly), mandibular prognathism, mandibular condylar deformation with hyperplasia of the coronoid process, bifidity of multiple ribs and giant multilocular odontogenic jaw cysts. Extensive multilamellar calcifications were found in falx cerebri, tentorium, falx cerebelli and in the apical segment of the atlanto-occipital ligament. Thoracic anomalies included bifid left 3rd, 4th, 5th and 6th rib, dismorphic body of the 3rd thoracic vertebra, dorsolumbar scoliosis, sacrum acutum. Interestingly, 3D-CT scan showed asymmetry of both sphenoid wings with thickening of the left wing sphenoid wing together with irregularity of the architecture of trabecular bone with alternating osteolytic and sclerotic areas. Abnormalities of the sphenoid bone are not very common, and consist of differently aggressive entities: some of them are typical of the pediatric age in few hereditary and congenital disorders.

The application of new criteria (i.e. peculiar calcifications of ligaments and sphenoid asymmetry) to a wider case series can lead to the early diagnosis of Gorlin syndrome, especially in pediatric patients, when the full phenotype is not yet expressed. The inclusion of bifid ribs as a novel major criteria and the recognition of peculiar cranial anomalies such as sphenoid asymmetry, well detected at volume CT reconstruction, might be useful for the recognition and characterization of misdiagnosed cases.