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Background
A minority of children with Osteogenesis Imperfecta (OI) seen within the Sheffield National Severe, Complex and Atypical Service (SCAOI) were also identified as showing symptoms consistent with an Autism Spectrum Disorder (ASD) (Balasubramanian *et al*. 2018). Diagnosis of ASD in conjunction with OI may be delayed due to presenting problems being inappropriately attributed to OI resulting in specialised ASD input not being received by children.
Presenting problem
We present two case examples of young children seen within the NHS England Severe and Complex Osteogenesis Imperfecta Service (SCAOI) who received additional diagnoses of ASD. These two children were aged 4 years and 6 years old at the time of diagnosis of ASD whereas they had received diagnoses of OI in infancy. Their early development was unusual, both displaying delayed language & communication. Apparent features that would be expected to be noticed in typical development included repetitive behaviours such as rocking/flapping and distinctly unusual communication styles and play. Neither child had been identified as potentially having ASD by community services such as health visitor checks or by nursery. Both children showed challenging behaviours.
Clinical management
Diagnosis of ASD has had significant positive effects for both children and their families. Professionals working with children with SCAOI need to be alert for ASD 'Red Flags'. This presentation details the diagnostic process, interventions and outcomes and includes Child A transitioning from mainstream to special school with the more supportive environment resulting in a significant decrease in problem behaviours. Child B remained in mainstream school but received specialist ASD support and the school benefited greatly from the understanding of the child in the context of their additional ASD diagnosis.
Discussion
Due to the possibility of dual diagnosis being overlooked as a result of the motor delay present with OI and a lack of understanding of OI outside of specialist services, it is very important for specialist services to notice ASD 'red flags' as detailed in this presentation and refer for full assessment where required.
Disclosure
NJB consults for Alexion, Mereo, UCB and Amgen, and receives grant support for clinical studies from Alexion and Amgen. PA receives Honoraria/expenses: Alexion and Kyowa Kirin and expenses from BioMarin.

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Hypercalcemia and parathyroid hormone-related peptide expression in a 3 months old boy with Colon Hemangioendothelioma

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Introduction
Epithelioid hemangioendothelioma (HEE) is a tumor of vascular origin, infrequent in the pediatric age and even more infrequent at intestinal level. To our knowledge, there are no previous reports of pediatric patients with malignant humoral hypercalcemia associated with this tumor. Humoral mechanism is seen more often in lung, uterine cérvix, skin and esophagus tumors. The presence of hypercalcemia appears to be an ominous prognostic sign.
Objective
To report the first case of a patient with hypercalcemia related to PTH-rP associated with Colon HEE in a pediatric patient.
Case description
A 3 months years old boy was admited because of clinical worsening and palpable abdominal mass. Initial laboratory investigation revealed hypercalcemia with the following workup: PTH: 1.65 pg/ml, calcium: 25.1 mg/dl, phosphorus 2.9 mg/dl 25ohvitamine D:25.2 ng/ml, Urine catecholamines were normal. Ultrasound visualized a highly vascularized tumor with calcifications in retroperitoneum of $8\times6\times6$ cm located between liver and right kidney. Biphosphonates and Calcitonin were iniciated without improvement. Biopsy reported epithelioid hemangioendothelioma and angiography revealed tumor irrigated by the middle colic artery. Selective embolization was performed with spongostan and 24 hours later, tumor exeresis was achieved. PTHrP mRNA was identified in the tumor. After surgery the patient attained normocalcemic state, PTH levels normalized and remained normocalcemic to date, 18 months later.
Conclusion
We report the first case of PTH-rp related hypercalcemia, with mesocolon epithelioid hemangioendothelioma a pediatric patient. PTH-rp mRNA was detected at tumor level, and the patient resolved hypercalcemia with tumor resection, remaining normocalcemic and with normal PTH levels since then. Selective embolization was important in order to facilitate tumor resection successfully, and improving morbidity and mortality of this surgery.
Disclosure
The authors declared no competing interests.

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Comparison of cell separation methods, using relative expression of specific growth plate zone markers in a pig model

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Objective
Linear growth is achieved by enchondral ossification in epiphyseal growth plates (GP) of long bones. These highly organized cartilaginous tissues contain chondrocytes of all differentiational stages classified in 3–5 specific zones. Due to their discrete characteristics, distinct analysis of each zone is essential in basic GP research. While the efficiency of zonal separation is therefore highly influencing on study results, comparative data on commonly used methods are sparse. This study aims to compare the efficiency of density gradient centrifugation (DGC) and laser capture microdissection (LCM) by quantitative real time PCR (qRT-PCR) of zone-specific growth plate samples.
Methods
Primary chondrocytes and cartilage tissues were isolated from femoral and tibial growth plates of prepubertal piglets and separated by density gradient centrifugation and Laser Microdissection (LCM) respectively. Samples were evaluated by qRT-PCR for Secreted Frizzled Related Protein 5 (Sfrp5) and Collagen type X (ColX) expression as markers for resting and hypertrophic zone, respectively.
Results
Significant differences in marker gene expression for resting and hypertrophic zones as compared with their respective adjacent zones could be found in both separation methods. Both LCM and density gradient centrifugation were able to discriminate resting vs. proliferative zones by Sfrp5 expression values, although to different levels of significance (DGC: $P=0.034$; LCM $=0.003$). Comparable results were observed for ColX gene expression levels in hypertrophic versus proliferative zones (DGC $P=0.024$; LCM <0.001).
Conclusion
While both methods are able to discriminate growth plate zones, LCM achieved a higher level of significance in zonal separation as compared to DGC. Thus, LCM allows to minimize methodical bias and should be the preferred method for expressional studies on specific growth plate zones.
Disclosure
The authors declared no competing interests.

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