assessment of deformity progression and changes in planned surgical management as a result of delay were recorded. Patients were graded on the degree of harm suffered as a consequence of delayed treatment utilising the National Patient Safety Agency harm criteria (United Kingdom).

Results: 94 patients were awaiting surgery for greater than 12 months (range 12 - 80). Of these 4 patients were graded as suffering severe harm including reduced pulmonary function. Average age at surgery was 7.8yrs (4-12.8). Average curve deterioration whilst awaiting surgery was 27 (14-43) degrees for major and 21 (8-56) degrees for minor curves. Postoperative correction averaged 37% (61-14%) for major and 48% (72-17%) for minor curves. Planned anterior surgery was abandoned in 1 patient and 1 patient underwent definitive posterior instrumented fusion instead of growing rods due to curve progression or worsening respiratory function. 2 patients suffered moderate harm requiring an unplanned anterior release due to delay. In all cases combined factors contributed to delays. Organisational delays along with composite delays in medical management across specialties significantly contributed to delayed surgery.

Conclusion: Delayed surgical intervention in EOS can result in severe patient harm. Greater integration of medical and surgical specialties with investment in organisational infrastructure could potentially reduce episodes of harm in our organisation.

Author disclosures: Morgan Jones: none. Matthew P Newton Ede: Stryker. Jwalant Metha: none. Adrian Gardner: none. Jonathan B Spilsbury: none. David Marks: Depuy Synthes; K2M; Medtronic; Stryker.

Paper #16

HIF-1α critical to Somitogenesis and interacts with the Notch pathway leading to vertebral malformations

Angela Yao, Frances Farley, Ernestina Schipani

Summary: HIF-1α is critical to somitogenesis. Lack of HIF-1α disrupts the Notch signaling pathway which leads to vertebral malformations seen in congenital scoliosis.

Hypothesis: To develop a knockout mouse model for Jarcho-Levin Syndrome using HIF-1α knock out mice.

Design: To produce and characterize a HIF-1α knockout mouse. We sought to characterize this mutant mouse and test the mutant mouse for common Notch pathway genes.

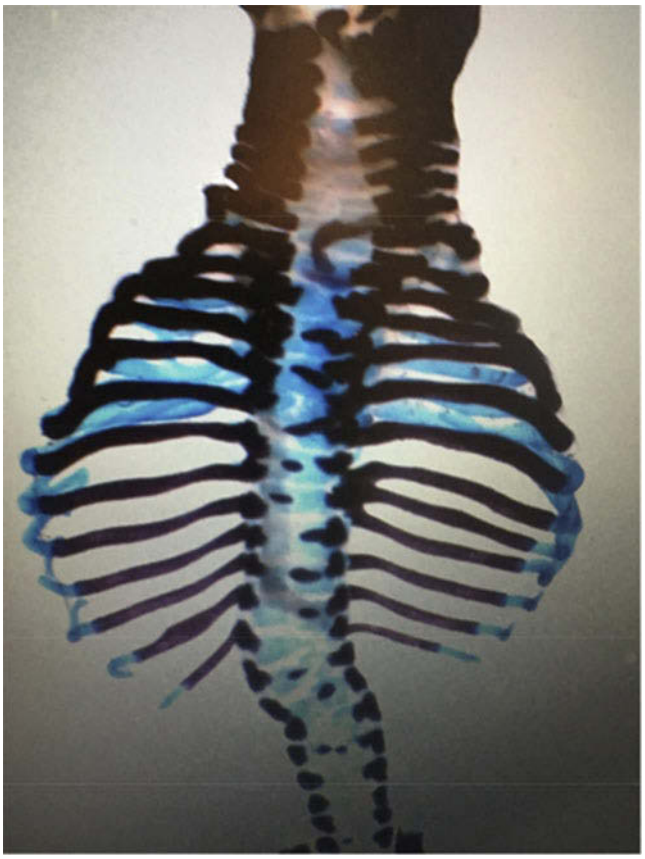
Introduction: HIF-1α is a gene that is up regulated in a hypoxic environment. Jarcho-Levin syndrome includes severe congenital scoliosis and is associated with 4 genes in the Notch pathway of somitogenesis.

Methods: A HIF-1α knockout mouse model was produced. The mutant phenotype was characterized. The mutant phenotype was tested to see whether the vertebral malformation was affected during somitogenesis versus endochondral bone development by conditional knocking out HIF-1α chondrocytes using Col2a1-Cre mouse line. The mutant HIF-1α-CKO embryos were tested for the expression of four Notch pathway genes (Dll1, Lfng, Hes7, and Mesp2).

Results: 25 HIF-1α CKO mutant mice were analysed from embryonic 14.5 days to Newborn. These mutant mice have many vertebral and rib abnormalities similar to those seen in Jarcho-Levin Syndrome. Conditional knocking out HIF-1α chondrocytes using Col2a1-Cre mouse line did not produce the vertebral abnormalities. 31 mutant embryos analyzed by whole mount in situ show an abnormal pattern of segmented somites, and there was disrupted expression of Dll1, Lfng, Hes7, and Mesp2 in the mutant embryos.

Conclusion: The HIF-1α-CKO mutant mice are a mouse model for Jarcho-Levin Syndrome. HIF-1α is important in somitogenesis. Notch signaling pathway is disrupted in the HIF-1α-CKO mutant mice.

Author disclosures: Angela Yao: none. Frances Farley: none. Ernestina Schipani: none.



Paper #17

Early Onset Scoliosis within the 22q11.2 Deletion Syndrome (22q11.2DS)

Jelle Homans, Vyaas Baldew, Tom Schlösser, Moyo Kruyt, René Castelein

Summary: 22q11.2DS is the most common microdeletion syndrome. It is characterized by a wide phenotypic variability, including scoliosis. This is the first epidemiological study that shows that the prevalence of early onset scoliosis within this vulnerable group of patients is around 40%. A congenital heart defect (CHD) irrespective of surgery was an independent risk factor, with an Odd′s Ratio of 12.09.

Hypothesis: The prevalence of early onset scoliosis within 22q11.2DS is increased

Design: Cross-sectional, based on prospectively collected data.

Introduction: 22q11.2DS is the most common microdeletion syndrome with a prevalence of 1:4000 new-borns. It is known to have wide phenotypic variability, including scoliosis, that, often, strongly resembles idiopathic scoliosis. The prevalence of scoliosis within 22q11.2DS is unknown. This epidemiological study on prospective data identifies the prevalence and clinical risk factors associated with the development of early onset scoliosis within 22q11.2DS.

Methods: Since 2014 all 22q11.2DS patients are radiographically screened for scoliosis in our 22q11.2DS clinic. All patients less than ten years old that visited the outpatient clinic between January 2014 and June 2017 were included. The prevalence of scoliosis (>10 degrees Cobb Angle) was determined. The criteria defined by Spiegel et al. (2003) were used to divide curves into ′typical′ (idiopathic-like) or ′atypical′ curves. Furthermore, clinical characteristics that may be associated with the presence of a scoliosis, like CHD with or without