POSTER PRESENTATIONS

P33

A REPORT OF TWO LIFE-BORN INFANTS, HOMOZYGOUS FOR SOUTHEAST ASIAN OVALOCYTOSIS

Surini Yusoff¹, Narazah Mohd Yusoff², Keng Wee Teik³, Chng Gaik Siew³, Winnie Ong Pei Tee³,
Yeoh Seoh Leng⁴, Nur Hasnah Ma’amor⁵, Masafumi Matsuo⁶, Hans Van Rostenberghe¹

1. Department of Paediatrics, Universiti Sains Malaysia
2. Advanced Medical and Dental Institute, Universiti Sains Malaysia
3. Department of Genetics, Hospital Kuala Lumpur, Kuala Lumpur
4. Department of Paediatrics, Hospital Pulau Pinang
5. Human Genome Centre, Universiti Sains Malaysia, Malaysia
6. Department of Paediatrics, Kobe Graduate School of Medicine, Kobe, Japan

Introduction: Homozygosity of Southeast Asian Ovalocytosis (SAO) has been thought to cause death in utero. Life births have not been reported so far. We report two cases of live-born infants with homozygous codon 400-408 deletion of the erythrocyte band 3 gene.

Case presentations: Probands 1 and 2 were boys, born to mothers aged 25 and 35 years respectively via emergency lower segment Caesarean section for foetal distress. Proband 1 was born with hydrops fetalis and presented with pallor, oedema, hepatosplenomegaly and a globular heart on chest X-ray. There was severe anaemia (Hb 4.9 g/dL) at birth with haemolytic picture on the blood film. The infant died at 7 hours of life. Proband 2 was born to a mother who had previously three spontaneous abortions. This case was diagnosed antenatally to have severe anaemia and received 2 antenatal transfusions. He was ventilated after birth for respiratory distress. Haemolytic anaemia complicated by pathological jaundice was ongoing and he required exchange transfusion at day 2 and a packed transfusion at day 5 of life. The infant was clinically bronzed with large hepatomegaly (4-5cm) and palpable spleen (1cm). The DNA analysis revealed that both probands were homozygous for the codon 400-408 deletion in band 3 gene and the parents were heterozygous.

Conclusion: This is the first report of two live-born infants with homozygosity of the band 3 gene (SAO). One of the children received antenatal treatment for foetal anaemia and managed to survive up to at least two months of age.