An Unusual Cause of Pancytopenia in a Patient with Respiratory Syncitial Virus Bronchiolitis

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A 4 week old baby presenting with RSV positive bronchiolitis was ventilated for 48 hours. Unexpectedly he had pancytopenia and hepatosplenomegaly. Laboratory results were inconsistent with haemolysis. A bone marrow aspirate obtained with difficulty, revealed a leuкоerythrobластic picture consistent with inflammatory response to infection. Several chest X-rays were done to optimise ventilation. The radiologist commented on a generalised increased bone density which prompted a skeletal survey that revealed features of osteopetrosis.

Osteopetrosis is a group of inherited conditions characterised by increased bone density [1]. Failure of osteoclastic bone resorption leads to abnormal bone marrow cavity formation, persistence of primary spongiosa and abnormal bone remodelling. This results in marrow failure, frequent fractures and compression of cranial nerves, notably the optic nerve [2].

The severe recessive form presents within 3 months with visual concerns, failure to thrive, recurrent viral respiratory infections, pancytopenia and hepatosplenomegaly. However this rare condition is often suspected on a surreptitiously performed X-ray [1].

Diagnosis is based on clinical features and skeletal survey. Several genes (CLCN7 gene [3], the TCIRG1 gene, OSTM1 genes, RANKL gene [4]), are implicated. Disease modifying treatment involves bone marrow transplantation, prioritising infants with some preserved vision. Successful engraftment is greatly influenced by the extent of HLA matching.

Bibliography