Case reports

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Congenital non-chylous pleural effusion with Down's syndrome

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SUMMARY The association of Down's syndrome with congenital non-chylous pleural effusion is described in a preterm infant.

Case report

The patient, a girl weighing 1.39 kg, was born at 33 weeks' gestation by emergency caesarian section for fetal distress. The features of Down's syndrome were noted at birth and the diagnosis confirmed on chromosome study ($2n=47,XX,+21$). She developed respiratory distress within minutes of delivery and required ventilation. Chest x ray revealed complete opacification of the right hemithorax with a small area of radiolucency representing the unexpanded right lung (figure). Straw coloured fluid totalling 43 ml was removed by needle aspiration. The baby was extubated at the age of 12 hours but over the next seven days 10 further pleural taps were required because of increasing respiratory distress. A total of 370 ml of fluid was removed. The final pleural aspiration was performed on the seventeenth day of life with the removal of 30 ml. The protein content of the fluid ranged from 23 to 26 g/l, it contained scanty leucocytes, and showed no growth on culture. Daily intravenous albumin infusions were administered for the first five days of life with an initial total plasma protein level of 36 g/l rising to 50 g/l by the tenth day of life. Seven days after the start of introduction of milk feeds the fluid remained straw coloured and contained no fat.

Discussion

Congenital pleural effusion is uncommon but well described in the neonate. Yancy and Spock,¹ in a review of 31 cases, found the majority to be chylothorax, with the pleural fluid initially chylous with a milky appearance or becoming so subsequently. The infants reported were mainly born at term after normal labours and deliveries. There was a preponderance of males and the effusions were

more commonly right sided. The occurrence of chylothorax postoperatively is usually attributed to the trauma of thoracotomy, but the pathogenesis of the congenital form is obscure.

The typical milky chyle usually appears after milk feeds have begun but in the case described here the pleural fluid remained straw coloured seven days after the start of milk feeding. The fluid is thus unlikely to have originated from the thoracic duct, but rather from impaired lymphatic drainage of the pleural space and its resolution due to the maturation or development of alternative lymphatic channels. Patients with Down's syndrome have been

FIGURE Chest x ray showing unexpanded right lung.

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shown to have lungs with a smaller alveolar surface area and therefore presumed smaller capillary surface area,² which may predispose to the delayed clearance of lung fluid. That the effusion was a transudate is borne out by the scanty leucocyte count and protein content lower than plasma protein level.

Standard textbooks do not report an association of pleural effusions and chromosomal abnormalities, though Chernick and Reed³ commented on the association of chylothorax and Turner's syndrome and Yoss and Lipsitz⁴ reported chylothorax occurring in two infants with Down's syndrome.

Repeated needle aspiration is likely to be successful in ultimately controlling the reaccumulation of fluid. The insertion of a chest drain is therefore probably not justified in view of the hazards of infection and possible potentiation of high protein fluid loss.

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Homozygosity in piebald trait

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SUMMARY A severely affected child born to consanguineous parents is interpreted as being a homozygote for the dominantly inherited piebald trait. The striking phenotypic difference between the parents and the child implies intermediate inheritance of this condition, and the family also illustrates that consanguinity should not always be taken to indicate genetic heterogeneity and recessive inheritance.

Mendel defined a dominant character as one with identical or almost identical expression in the heterozygote and the homozygote.¹ A relevant comparison may be difficult in man as matings between heterozygotes may be rare and, in addition, a difference in phenotype may remain undetected because of early homozygote lethality. Thus, many human conditions classified as dominant may in fact be intermediately inherited, but there are comparatively few examples where heterozygotes have been

differentiated from the homozygote.²⁻⁴ We present here an Asian family with partial and complete albinism illustrating these points.

Case report

The proband and his family came from north-east Pakistan. He was born at term and weighed 3.2 kg. At birth he was noted to be an albino, but no other neonatal problems were reported. Over the subsequent months there was concern about his development, in particular his lack of response to sound and his slow motor development. He was admitted to hospital with a suspected febrile fit while visiting relatives in the UK at nine months of age. His growth was satisfactory (weight 10th to 25th centile, length 50th centile, head circumference 25th centile). He was hypotonic with poor head control, could not sit unsupported, and still possessed a primitive grasp reflex. He made no response to sound, hardly vocalised, and was considered totally deaf by his relatives. An EEG showed lack of physiological activity suggesting severe brain impairment. Apart from complete absence of hair and skin pigmentation and blue irides he showed some facial