Discussion

Persistent pulmonary hypertension in the newborn has a multiple aetiology and occurs in a variety of neonatal disorders. There is considerable evidence that thromboxane A2 is involved in at least some forms of pulmonary hypertension. Infusion of live group B β haemolytic streptococcus, streptococcal toxin, or purified *Escherichia coli* endotoxin in animal models causes pulmonary hypertension, which is associated with high plasma thromboxane B2 concentrations. It has also been suggested that pulmonary hypertension seen in perinatal disorders is mediated by release of platelet thromboxane A2 in the lungs. Inhaled amniotic fluid may be absorbed by the lungs and cause local platelet aggregation. Thromboxane A2 is then released from the platelets and causes pulmonary vasoconstriction.

Our results suggest that production of platelet thromboxane A2 is decreased in persistent pulmonary hypertension of the newborn. The lowest values were found in cases of severe meconium aspiration syndrome, but no correlations between the simultaneous blood gas values and production of platelet thromboxane B2 were found. Although samples in the two groups were drawn from different sites, this should not influence the thromboxane B2 results because they were expressed as nanograms per number of platelets. The high plasma thromboxane B2 concentration in previous studies may originate either from the lungs or from hyper-reactive platelets, which release thromboxane B2 excessively in vivo. If this in vivo stimulation of the release of thromboxane B2 lasts long enough it may lead to exhaustion of the capacity of the platelets to produce thromboxane B2. The present in vitro data on reduced production of platelet thromboxane B2 may support this hypothesis.

Vasoactive prostanoids and their inhibitors may be useful in the treatment of some forms of pulmonary hypertension. Selective inhibition of thromboxane A2 formation is effective in some examples of experimental pulmonary hypertension. The biological antagonist of thromboxane A2, prostacyclin, has been successfully used to reduce pulmonary hypertension. The response to this treatment with dilator could be enhanced if the platelet formation of the vasoconstrictive thromboxane A2 is already diminished, as in the present cases.

References


Correspondence to Dr P Kääpä, Department of Paediatrics, Central Hospital of North Karelia, SF-80210 Joensuu, Finland. Received 22 August 1986

Noonan’s syndrome and neurofibromatosis

A SHUPER, M MUKAMEL, M MIMOUNI, AND R STEINHERZ

Department of Pediatrics B, Biochemical and Developmental Genetic Unit, and Department of Pediatrics A, Beilinson Medical Center, Petach Tikva, and Sackler School of Medicine, Tel Aviv University, Israel

Noonan’s syndrome and multiple café au lait spots, compatible in size and number with von Recklinghausen’s neurofibromatosis, is presented. These features may represent a distinct genetic entity rather than the coincidence of two diseases.

Von Recklinghausen’s neurofibromatosis and

SUMMARY A child with Noonan syndrome and multiple café au lait spots, compatible in size and number with von Recklinghausen’s neurofibromatosis, is presented. These features may represent a distinct genetic entity rather than the coincidence of two diseases.
syndrome.\textsuperscript{4-6} We present one more case of this latter syndrome. Such cases have not been reported previously in paediatric publications.

\textbf{Case report}

The patient was referred to our clinic at the age of 12 years because of severe headaches. He was the fourth son of unrelated parents of Moroccan origin, both aged 47 years. The parents and the three siblings were all healthy, having normal height and weight and unremarkable physical findings. Pregnancy and delivery had been normal and birth weight had been 3500 g. Multiple cafe au lait spots were noted at birth and later were found to have increased in size and number. He sat up at the age of 9 months and walked alone at 18 months. No developmental problems were noted until he was of school age, when severe learning difficulties became apparent.

On examination, his weight was 30 kg (12th centile), height 132 cm (<3rd centile), and head circumference 55-5 cm (75th centile). There was bilateral ptosis of the eyelids, with a downward palpebral slant and low set ears (Figure (a)). The palate was high arched and the teeth overcrowded. His hair was coarse with a low posterior hairline. The neck was short with prominent pterygium colli. He also had pectus excavatum and cubitus valgus. Physical examination of the heart yielded normal results. The testicles were in the scrotum, with a volume of 2 ml. No pubic or axillary hair was present. Scattered over the entire skin surface were more than 20 cafe au lait spots (Figure (b)), with a diameter ranging from several millimetres to 10 cm. Of these, 10 were more than 1-5 cm in diameter. No fibromata could be found. Electrocardiography and echocardiography yielded normal results. Roentgenographic survey of the skeleton failed to show any bone anomaly aside from the pectus excavatum and cubitus valgus. His bone age was 7 years. The karyotype was normal. Computed tomography of the brain, performed because of the severe headaches, did not show any disease.

\begin{figure}[h]
\centering
\includegraphics[width=\textwidth]{image.png}
\caption{(a) Frontal face photograph of the child (left); (b) general appearance of the child (right).}
\end{figure}

\textit{Noonan's syndrome and neurofibromatosis} 197
Hydrops fetalis due to abnormal lymphatics

K P WINDEBANK, N A BRIDGES, I OSTMAN-SMITH, AND J E STEVENS

Department of Paediatrics, John Radcliffe Hospital, Oxford

SUMMARY A case of generalised lymphatic abnormality that presented with hydrops fetalis is described. This seems to be the first such case reported.

The reported incidence of non-immunological hydrops fetalis varies from one in 14001 to one in 7143.2 Causes are identified in 56%3 to 85%1 of cases, among which cardiac arrhythmia, thalassaemia, twin to twin transfusion, and congenital heart disease account for half.3 Localised lymphatic abnormalities—that is, cystic hygroma, pulmonary lymphangectasia,2 4 and prolonged chylos ascites2 —may be causes, but there are no reports of generalised lymphatic abnormalities being associated with hydrops.
Noonan's syndrome and neurofibromatosis.

A Shuper, M Mukamel, M Mimouni and R Steinherz

Arch Dis Child 1987 62: 196-198
doi: 10.1136/adc.62.2.196

Email alerting service

These include:

Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/