At birth, the proband (III 4) had petechiae and a platelet count of 14000/mm³ (fig 1). A bone marrow examination showed that the megakaryocytes were severely reduced in number. The child had also widespread eczema from the first few days of life. The child had first been admitted at the age of 2 months: his platelet count has been 12000/mm³, with an eosinophil count of 800/mm³, an IgG of 95 mg/dl, an IgM of 200 ml/dl, an IgA of 144 mg/dl, and an IgE of 2132 U/ml. Lymphocyte numbers, CD4:CD8 ratio, glucose tolerance, urinary acids and karyotypes were all normal.

Soon after admission he developed diarrhoea which did not respond to total parenteral nutrition. At the age of 3 months, biochemical signs of parenchymal renal disease were detected. The parents refused further treatment and the child died shortly afterwards.

III 1 died at the age of 6 months as a result of intractable diarrhoea. Total parenteral nutrition led to glucose intolerance. At the age of 1 month III 3 developed insulin dependent diabetes mellitus and diarrhoea; an intestinal biopsy specimen showed severe jejunal mucosal infiltration associated with infiltration of the lamina propria by lymphocytes and plasma cells. Autoimmune enteropathy was subsequently diagnosed. Despite treatment with steroids, cyclosporin, and total parenteral nutrition, he died at the age of 7 months as a result of hepatic damage and septic complications.

III 7 had had diarrhoea from the age of 45 days and normal glucose tolerance. Serum enterocyte autoantibodies and thyroid microsomal autoantibodies were detected and he was treated with cyclosporin, steroids, and total parenteral nutrition. He died at the age of 12 months.

III 8 had intractable diarrhoea and died at the age of 2 months.

III 9 had diabetes and died at the age of 4 months.

III 11 developed coeliac disease and is following a gluten free diet at the time of writing. He remains well at the age of 7 years.

The thrombocytopenia is a feature of another X linked disease—Wiskott-Aldrich syndrome. Thrombocytopenia, liver and renal diseases are rare in the families with neonatal diabetes and diarrhoea reported to date.

The case reported by Peake had agenesis of the islet of Langerhans; our patient had neonatal amegakaryocytic thrombocytopenia. These two features are not associated with immune dysregulation, which is probably not the primary defect.

Our family is interesting because of the different associations in each child, a picture which is not dissimilar to mitochondrial disease, but we have no other evidence in support of this hypothesis. It is our belief, however, that all the patients with X linked neonatal diabetes and diarrhoea reported to date, have, in fact, one disease only.

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Pain response in neonates

EDITOR—Dr Ramenghi and colleagues have elegantly shown that intranasal administration of sweet tasting solutions reduces the response to pain in the newborn. 1 2 Although young infants have been the focus of study so far, the findings may be applicable to older children. According to the following extract, this already seems to have been recognised some time ago. Charlotte Bronte published her novel Villette in 1853, based on her experiences of studying and working in Brussels. The character of Lucy Snowes relates what happened when the child of her employer broke an arm and was attended by the doctor, the bones being successfully reset once the child had been calmed with sugar water:

"I, at least, was taken up with endeavouring to soothe the Fifine; whose cries (for she had good lungs) were appalling to hear...So now, when we get a little calmer, we must commence business; and we will soon have that unlucky little arm bandaged and in right order. Hereupon he called for a glass of eau sucrée, fed her with some teaspoonful of the sweet liquid...promised her more when the operation should be over, and promptly went to work.; much as he had hurt her, she held out her hand to bid him a friendly good-night." 3

Lucy, the shrewd observer of this scene, commented that "the little patient's heart had been won through her palate." This seems a more poetic way of saying that the antinociceptive effect of sucrose is mediated through endogenous opiate release.

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Erratum

Fetal growth velocity: kinetic, clinical, and biological aspects.


For "anoxia" on page F14, lines 12 and 15, please substitute "damage". Interestingly, this error arose because the original manuscript from Italy used the expression "a nona" (an injury or damage).
Pain response in neonates.

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Arch Dis Child Fetal Neonatal Ed 1996 75: F144
doi: 10.1136/fn.75.2.F144-a

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