The Journal of Laryngology and Otology
October 1999, Vol. **113,** pp. 919–921

Feingold syndrome – a cause of profound deafness

ANNABEL DODDS, M.B., CH.B., M.SC.*, RICHARD RAMSDEN, F.R.C.S.†,
HELEN KINGSTON, M.D., F.R.C.P., D.C.H.‡

Abstract
A case of Feingold syndrome is presented with a previously undescribed association of bilateral profound hearing impairment. Computed tomography (CT) scanning revealed severe narrowing of the internal auditory meatuses at the peripheral end with non-existent auditory nerves. This pathology is significant in the future habilitation of the child, as cochlear implantation is not possible. Children with multiple congenital anomalies should have radiological investigations before hopes are raised with respect to cochlear implantation.

Key words: Deafness; Cochlear implant

Introduction

Feingold syndrome was first described in 1975 (Feingold) in a child who had microcephaly with normal development, mild facial dysmorphism, hand and foot anomalies, tracheo-oesophageal fistula and duodenal atresia. Identical hand abnormalities and microcephaly with normal intelligence in the patient's father and paternal grandmother suggested autosomal dominant inheritance. Additional cases providing further delineation of this rare syndrome have been reported (Feingold, 1978; König *et al.*, 1990; Brunner and Winter, 1991). A total of 23 cases have been reported although full clinical details are not available on all.

Microcephaly with normal intelligence or mild learning difficulties is a hallmark of the condition, together with characteristic digital abnormalities consisting of bilateral fifth finger clinodactyly (short curved finger) with additional mesobrachyphalangy (short middle phalanx) of the second finger in some cases and 2/3 and 4/5 syndactyly (skin fusion) of the toes. Short palpebral fissures are reported in most cases. Tracheo-oesophageal fistula/atresia occurred in six of the 23 cases and duodenal obstruction due to atresia, annular pancreas or duodenal membrane in five of the 23 cases. Additional features reported in single cases included the presence of multiple haemangiomata, congenital heart disease (tricuspid atresia with ventricular septal defect (VSD) and interrupted aortic arch) and one case of unilateral sensorineural deafness. The distinctive combination of features reported suggests a clinically recognizable, fully penetrant but variably expressed syndrome with autosomal dominant transmission.

Case report

We report a female born to a healthy unrelated Caucasian couple. A previous pregnancy had been terminated because of severe skeletal abnormalities affecting the limbs detected by ultrasonography. Polyhydramnios and breech presentation were noted during the pregnancy. Premature onset of labour occurred at 28 weeks gestation and delivery was by caesarean section for foetal distress. Birth weight was 960 gms. Resuscitation was required after delivery with ventilation for 24 hours.

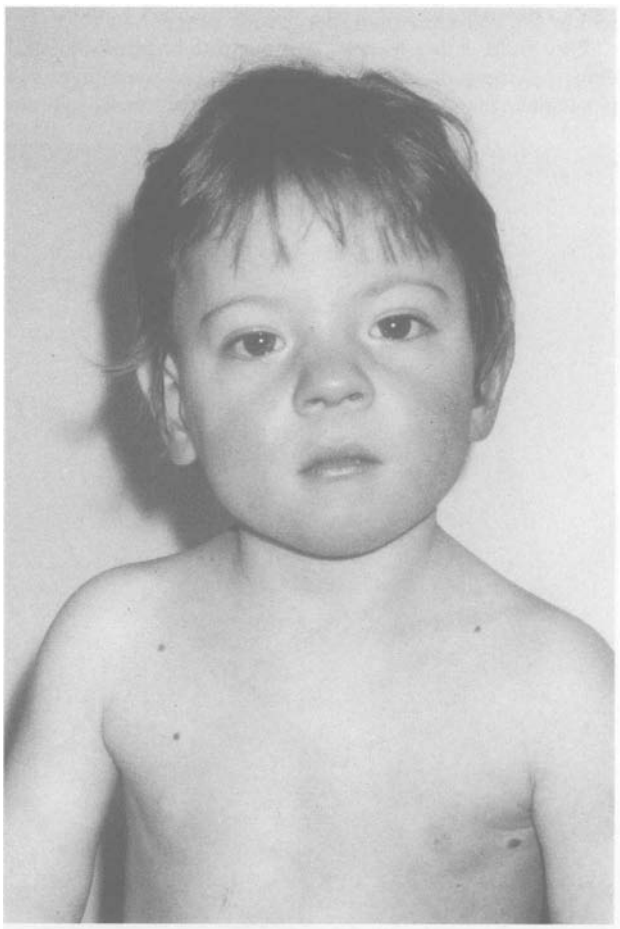

FIG. 1
Microcephaly, telecanthus, narrow palpebral fissures and multiple small cavernous haemangiomata.

From the East Lancashire Paediatric Audiology Service*, Nelson, the Department of Otolaryngology†, Manchester Royal Infirmary, Manchester, and the Regional Genetic Service‡, St Mary's Hospital, Manchester, UK.
Accepted for publication: 21 May 1999.

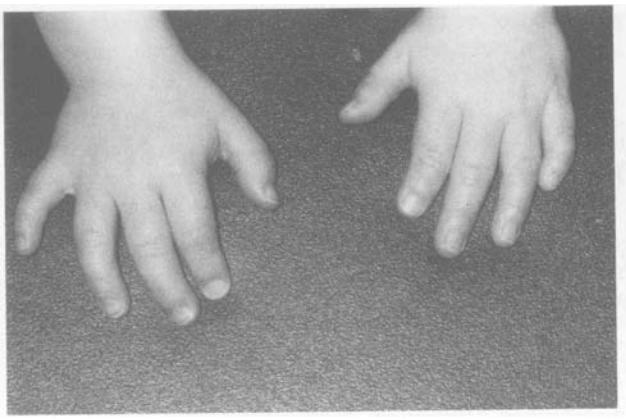

FIG. 2
Clinodactyly of index and fifth finger.

Duodenal obstruction was diagnosed following a barium study and duodenostomy for annular pancreas was performed on day five. The post-operative period was complicated by prolonged metabolic acidosis, necrotizing enterocolitis and staphylococcal septicaemia. Reventilation was required for 19 days because of recurrent apnoea and bradycardia. A patent ductus arteriosus was detected and treated with indomethacin on days 14 and 23. Intravenous gentamicin was administered but serum levels remained within the recommended therapeutic range.

The child subsequently demonstrated developmental delay affecting motor skills and language development in particular. Increased tone in the legs with brisk reflexes

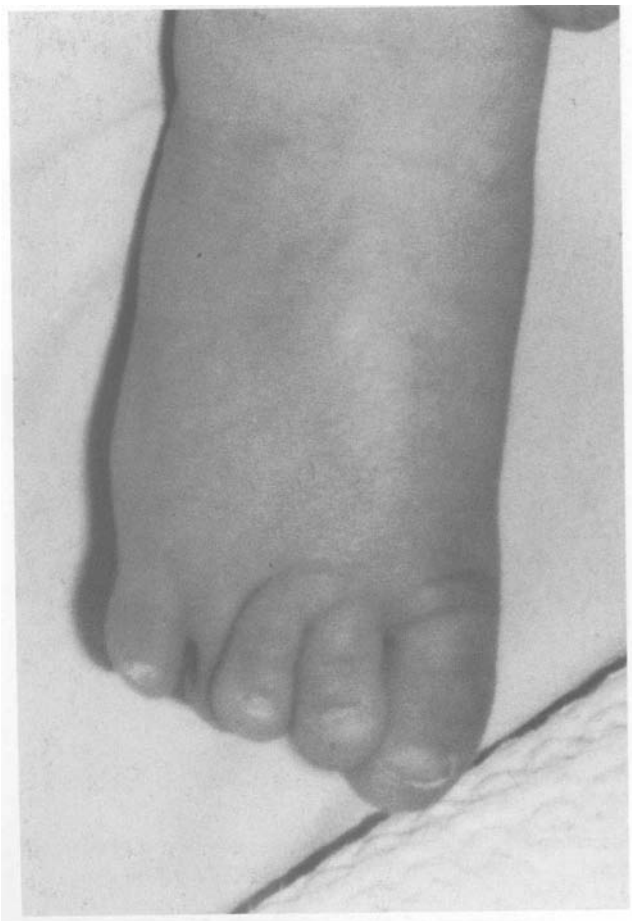

FIG. 3
Partial 2/3 and 3/4 syndactyly of the toes.

suggested the possibility of an emerging cerebral palsy. CT scan was normal apart from mild dilatation of the central and peripheral CSF spaces. Head circumference was 43.6 cm at three years and eight months of age, representing severe microcephaly in comparison to height and weight measurements on the 0.4th centile. A diagnosis of Feingold syndrome was made on the basis of the accompanying dysmorphic features comprising clinodactyly of the index and fifth fingers with shortening of the middle phalanges, partial 2/3 and 3/4 syndactyly of the toes, telecanthus (lateral displacement of the inner canthi) with short palpebral fissures and multiple small cavernous haemangiomata (Figures 1–3). Skeletal survey and chromosomal analysis were normal and there was no increased chromosomal instability demonstrated in mitomycin C stressed cultures. The mother's head circumference was on the 10th centile and father's on the 50th centile. Neither parent demonstrated any other dysmorphic features. Neonatal screening for hearing impairment using auditory brainstem response (ABR) identified a profound hearing impairment. At three years of age very little speech had developed and communication was mainly through signing. At the age of four years she was gaining no benefit from very high-powered hearing aids and cochlear implantation was considered. A CT scan was carried out prior to referral to the cochlear implant team. This showed a very rare anomaly of severe narrowing of the internal auditory meatuses bilaterally (Figure 4). The meatuses were felt to be so narrow that they could not accommodate more than one cranial nerve and they were seen to be continuous with, and the same diameter as the labyrinthine portion of the facial nerve. Additionally as the child had normal facial function on both sides it was felt that there could not be a cochlear nerve present. Consideration was given to carrying out magnetic resonance imaging (MRI) to confirm the pathological findings but after consultation with the family who were resistant to any further investigation it was decided not to proceed. Future habilitation is concentrating on developing manual communication skills, as the child will not benefit from conventional hearing aids or a cochlear implant.

Discussion

The diagnosis of Feingold syndrome in this child was considered because of the dysmorphic features present in combination with microcephaly and duodenal obstruction due to an annular pancreas. The diagnosis of this

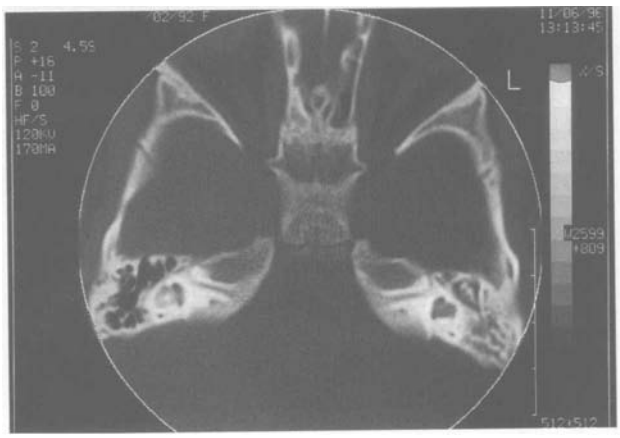

FIG. 4
Axial CT scan of temporal bones – the internal auditory meatuses are very narrow and are continuous with, and the same diameter as the labyrinthine portion of the facial canal. Note the inner dysplasia of the left inner ear.

https://doi.org/10.1017/S0022215100145591 Published online by Cambridge University Press

syndrome is based on the overall pattern of anomalies, that is quite distinct. Similar physical features can occur in deletions of chromosome 13 distal to band 13q14 (Schinzel, 1984), but the normal karyotype in this case excluded this possibility. In this family, neither parent was affected suggesting that the disorder in this case arose by new mutation. The hearing loss was assumed to be due to the perinatal factors – prematurity, low birth weight, ventilation, septicaemia, gentamicin administration and metabolic acidosis, all of which are high risk factors. The more severe degree of microcephaly and developmental delay present compared to previously reported cases may be explained by the prematurity and neonatal complications that occurred and the profound sensorineural hearing loss in our case. Unilateral total hearing loss has been described previously in one patient with Feingold syndrome (Brunner and Winter, 1991) but has not been investigated radiologically. Radiological investigation in the case described was carried out to assess the cochleas with a view to cochlear implantation. The cochleas were normal on CT scan but severe narrowing of the internal auditory meatuses excluded the child as a candidate for cochlear implantation. A narrow internal auditory canal will most likely contain only the facial nerve and in bilateral cases is a contraindication to cochlear implantation (Phelps, 1998). Magnetic resonance imaging (MRI) can demonstrate the nerves passing into the inner ear and should now be carried out on all potential cochlear implant candidates if CT scanning has demonstrated abnormal pathology of the inner ear or internal auditory meatus. Similar pathology to the case previously described with unilateral total hearing loss cannot be excluded. Audiologically, the importance of this case study is the outcome with regard to the future habilitation of the patient. Cochlear implantation is now considered for children with profound congenital hearing loss as young as 12 months and this case illustrates the importance of CT scanning at an early stage, especially in children with other congenital anomalies. As a result of the increasing expertise in CT scanning of the auditory system it is likely that many more anomalies will be identified over the next few years. Audiologists, otologists and paediatricians must look for these anomalies and must consider middle-ear, inner-ear and temporal bone anomalies in children with hearing loss in which the clinical picture is unclear or 'does not fit'. The clinician still needs to evaluate the possible benefits of CT scanning carefully as very young children will require sedation to obtain clear images of the auditory apparatus. However, when children are being considered for cochlear implantation and have additional congenital anomalies it is wise to refer for a CT scan at an early stage and not raise false hopes. With the current views on neonatal screening focusing on otoacoustic emission testing audiologists have to be aware that, although the majority of congenital losses are due to pathology of the cochlea, retro-cochlear pathology does exist. There will, therefore, be implications for ongoing surveillance if otoacoustic emission is the preferred test in neonatal screening for hearing impairment.

References

Brunner, H. G., Winter, R. M. (1991) Autosomal dominant inheritance of abnormalities of the hands and feet with short palpebral fissures, variable microcephaly with learning disability, and oesophageal/duodenal atresia. *Journal of Medical Genetics* **28:** 389–394.

Feingold, M. (1975) Case report 30. *Syndrome Identification* **3(1):** 16–17.

Feingold, M. (1978) An unusual microcephaly. *Hospital Practice* **13:** 44–49.

König, R., Selzer, G., Stolp A., Fuchs, S. (1990) Microcephaly, mesobrachyphalangy and tracheoesophageal fistula: MMT syndrome. *Dysmorphology and Clinical Genetics* **4(3):** 83–86.

Schinzel, A. (1984) Chromosome 13 deletions. In *Catalogue of Unbalanced Chromosome Aberrations in Man*. De Gruyter, Berlin, pp 485–489.

Phelps, P. (1998) Imaging for cochlear implants: the importance of the latest techniques for pre-operative assessment. *Journal of Audiological Medicine* **7(3):** 145–154.

Address for correspondence:
Dr Annabel A. Dodds, M.B.Ch.B., M.Sc.,
Consultant Community Paediatrician in Audiology,
East Lancashire Paediatric Audiology Service,
Audiology Clinic,
Leeds Road,
Nelson BB9 9TG.