mechanisms: (a) presence of an occluding membrane that subsequently ruptured; (b) presence of an oesophageal fold working as a valve that became incompetent with ageing;[3] (c) progressive slope of the fistulous tract due to the different growth rates for the trachea and oesophagus.[6]

The location of the fistulous opening within the airway is usually the carina (91%).[7] Location in the right main bronchus is observed in 8% of patients. A congenital fistula between the oesophagus and left main bronchus has not been reported in the medical literature.

The diagnostic techniques are a barium swallow and bronchoscopy. In our patient bronchoscopy allowed identification of the fistulous opening. The sensitivity of both diagnostic methods is summarised in the table. Oesophagoscopy has a lower yield than the other two procedures as it can be very difficult to visualise the oesophageal opening. This should always be performed, however, to rule out other causes.[9,10] In some cases, including this one, the orifice of the fistula is hard to identify and hence a Nissen funduplication for gastro-oesophageal reflux is performed. The persistence of the symptoms after surgery led to a repeat barium swallow which demonstrated the lesion.

Percentage accuracy of bronchoscopy and barium swallow in the diagnosis of tracheo-oesophageal fistula as reported by different authors

	Bronchoscopy	*Barium swallow*
Bedard *et al*[1]	66	53
Kirk and Dicks-Mireaux[2]	100	47
Vasquez *et al*[3]	25	100
Johnston and Hastings[7]	50	66
Beaseley and Myers[8]	–	73
Mean	60	68

1 Bedard P, Girvan DP, Shandling B. Congenital H-type tracheoesophageal fistula. *J Pediatr Surg* 1974;**9**:663–8.
2 Kirk JME, Dicks-Mireaux C. Difficulties in diagnosis of congenital H-type tracheo-oesophageal fistulae. *Clin Radiol* 1989;**40**:150–3.
3 Vasquez RE, Landay M, Kilman WJ, Estern A, Schreiber T. Bening esophagorespiratory fistulas in adults. *Radiology* 1988;**167**:93–6.
4 Blackburn WR, Amoury RA. Congenital esophagopulmonary fistulas without esophageal atresia. An analysis of 260 fistulas in infants, children and adults. *Rev Surg* 1966;**23**:153–75.
5 Sheiner NM, Lachance C. Congenital esophagobronchial fistula in the adult. *Can J Surg* 1980;**23**:489–90.
6 Demong CV, Grow JB, Heitzman GC. Congenital tracheoesophageal fistula without atresia of the esophagus. *Am Surg* 1959;**25**:156–62.
7 Johnston PW, Hastings N. Congenital tracheoesophageal fistula without esophageal atresia. *Am J Surg* 1966;**112**:233–40.
8 Beaseley SW, Myers NA. The diagnosis of congenital tracheoesophageal fistula. *J Pediatr Surg* 1988;**23**:415–7.
9 Black RJ. Congenital atresia tracheo-oesophageal fistula in the adult. *Thorax* 1982;**37**:61–3.
10 Peat SK, Paes T, Howard ER, Jones PIE. Congenital tracheo-oesophageal fistula in an adult. *Br J Surg* 1989;**76**:412.

Thorax 1994;**49**:836–837

Congenital cutis laxa with a dominant inheritance and early onset emphysema

E Corbett, H Glaisyer, C Chan, B Madden, A Khaghani, M Yacoub

Abstract

Two cases (mother and daughter) are reported of autosomal dominant cutis laxa which are unusual in being associated with early onset emphysema. Both mother and daughter have been smokers and are heterozygotes for the α_1 antitrypsin genotype. The combination of cigarette smoking and subnormal α_1 antitrypsin levels may explain the pulmonary spread in these two women who have what is usually a benign form of cutis laxa limited to the skin.

(*Thorax* 1994;**49**:836–837)

Harefield Hospital, Harefield, Middlesex WB9 6JH
E Corbett
H Glaisyer
C Chan
B Madden
A Khaghani
M Yacoub

Reprint requests to:
Dr H Glaisyer,
26 Terrick Road,
Wood Green, London
N22 4SH.

Received 4 March 1992
Returned to authors
29 April 1992
Revised version received
23 July 1992
Accepted for publication
12 August 1992

Case reports

CASE 1

A 51 year old woman had been noted at birth to have an abnormal "velvety" skin and had developed the typical features of cutis laxa in her early childhood. By the age of 51 she looked much older than her chronological age. Her skin was lax and inelastic with redundant folds present, particularly on the face, neck, and abdominal wall. Her ear lobes were pendulous and the eyelids and nasolabial folds drooping despite cosmetic surgery to the face in her late teens. Her family history revealed a similarly affected mother and grandmother, both of whom were non-smokers who survived into their eighth decade. There was no family history of respiratory disease or consanguinity.

The patient became progressively dyspnoeic on exercise in her late twenties and was diagnosed as having emphysema. She had smoked 10–15 cigarettes a day from her early teens until diagnosed. Her symptoms progressed until July 1991 when she received a double lung transplant. Pretransplantation spirometry results are given in the table, and the appearance of the chest radiograph before transplantation is shown in the figure. Histopathological examination of the explanted lungs showed extensive emphysema with a scarred and bronchiectatic lingula. She is currently well 12 months after surgery with a forced expiratory volume in one second (FEV_1) of 2·0 l and forced vital capacity (FVC) of 2·3 l.

Pretransplantation investigations revealed a low serum α_1 antitrypsin level of 1·65 mg/l (normal ranges 1·6–2·1 mg/l) and a PI^{mz} genotype. Her serum IgG and IgM levels were also below the normal range at 4·6 g/l (normal range: 6·4–16) and 0·3 g/l (normal range: 0·6–2·8), respectively.

*Respiratory function values for cases 1 and 2**

	Case 1		Case 2	
	Actual	*% Predicted*	*Actual*	*% Predicted*
FEV_1 (l)	0·36	17	2·04	62
FVC (l)	1·10	39	4·40	107
FEV_1/FVC	32	44	46·4	57
PEF (l/min)	119	33	260	58
V_{50} (l/s)			1·14	23
V_{25} (l/s)			0·53	18
TLC (l)			7·20	131
RV (l)			2·46	173
RV/TLC (%)			34·2	142
TLCO (mmol/min/kPa)			8·73	92
KCO (mmol/min/kPa/l)			1·11	58

FEV_1 = forced expiratory volume in one second; FVC = forced vital capacity; PEF = peak expiratory flow rate; V_{50} = forced expiratory flow rate at 50% vital capacity; V_{25} = forced expiratory flow rate at 25% vital capacity; TLC = total lung capacity (determined by closed circuit helium dilution method); RV = residual volume (determined by closed circuit helium dilution method); TLCO = carbon monoxide transfer factor; KCO = transfer coefficient (determined by single breath method).
*Predicted values from Cotes JE, ed. *Lung function assessment and application in medicine*. 3rd edn. Oxford: Blackwell, 1975.

CASE 2

The 23 year old daughter of case 1 also had abnormal skin texture at birth and a loose, pendulous skin from early childhood. She is a current smoker of 15–20 cigarettes a day, having started in her late teens. There is no history of wheeze or atopy but she does have mild dyspnoea on exercise.

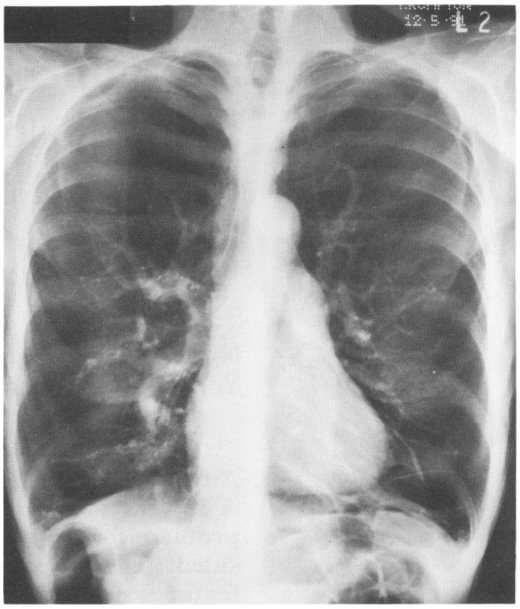
Despite the family history she has so far declined radiological investigations and follow up, but has had one set of lung function tests (table), the results of which are compatible with early emphysema. Her serum α_1 antitrypsin level is 1·02 mg/l with a PI^{mz} genotype and her immunoglobulin levels are within the normal range.

Discussion

Cutis laxa comprises a heterogeneous group of congenital and acquired conditions characterised by a distinctive lax and inelastic skin. The clinical features of cutis laxa are thought to result from abnormalities of elastin synthesis which result in the sparse and abnormal elastic fibres seen in affected tissues.[1–4]

The congenital variety is inherited either as a severe recessive condition with multisystem involvement,[5,6] or as an X linked condition with musculoskeletal and genitourinary abnormalities,[3] or as an autosomal dominant condition.[7]

Pulmonary involvement with early onset emphysema is a feature of the autosomal recessive and acquired forms of cutis laxa[5,8] but is unusual in patients with the autosomal dominant form. The mother and daughter reported here both have cutis laxa and an inheritance pattern that is consistent with the autosomal dominant form. The mother has emphysema that was clinically apparent at the age of 32 years. Her symptoms progressed to the extent that she was referred for lung transplantation at the age of 51 years. Her daughter's lung function test results suggest that she may follow a similar clinical course to that of her mother.



Posteroanterior chest radiograph of case 1 before lung transplantation showing hyperexpanded lung fields.

These two cases could represent a form of dominantly inherited cutis laxa in which pulmonary involvement is a prominent early feature of the syndrome. The previous two affected individuals in this family, both of whom were non-smokers, had a life span of more than 70 years with no significant respiratory symptoms. While it is conceivable that the phenotypic variation within this family represents variable penetration or anticipation, we feel that it is more likely that the respiratory involvement in the two individuals reported here results from a combination of congenitally abnormal elastin and the effects of cigarette smoke, possibly exacerbated by low α_1 antitrypsin levels. We suggest that individuals with the rare autosomal dominant form of cutis laxa should be advised of the special risks of cigarette smoking, especially if they are α_1 antitrypsin heterozygotes.

1 Goltz RW, Hult AM, Goldfarb M, Gorlin RJ. Cutis laxa: a manifestation of generalised elastolysis. *Arch Dermatol* 1965;**92**:373–87.
2 Oku T, Nakayama E, Imaizumi S, Takigawa M, Yamada M. Congenital cutis laxa. *Dermatologie* 1989;**179**:1983–7.
3 Byers PH, Siegel RC, Karen MD, Holdbrook A, Narayanari AS, Bornston P, *et al*. X-linked cutis laxa. Defective cross link formation in collagen due to decreased lysyl oxidase activity. *N Engl J Med* 1980;**303**:61–5.
4 Nanko H, Jepsen LV, Zachariah H, Sogaard H. Acquired cutis laxa (generalised elastolysis): light and electron microscopic studies. *Acta Derm Venerol (Stockh)* 1979;**59**:315–24.
5 Christeans L, Marchant-Alphant A, Forest M. Emphyseme congenital et cutis laxa. *Presse Med* 1954;**62**:1799–801.
6 Allanson J, Austin W, Hecht F. Congenital cutis laxa with retardation of growth and motor development: a recessive disorder of connective tissue with male lethality. *Clin Genet* 1986;**29**:133–6.
7 Beighton P. The dominant and recessive forms of cutis laxa. *J Med Genet* 1972;**9**:216–21.
8 Turner-Stokes L., Turton C, Pope FM, Green M. Emphysema and cutis laxa. *Thorax* 1983;**38**:790–5.