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ORIGINAL STUDY

Facial features in children with idiopathic congenital talipes equinovarus

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We assessed whether there is a recognisable pattern of facial morphology in photographs of a series of 62 children with idiopathic congenital talipes equinovarus (CTEV).

Photographs were scored for a number of facial characteristics by a research team comprising orthopaedic surgeons and clinical geneticists, to identify a subgroup of children with idiopathic CTEV, who shared characteristic facial features.

Seven children were identified as having a "CTEV face". There was good correlation between the "CTEV face" and the individual facial features. Repeatability of the assessment was good, and there was good correlation between the geneticists and the orthopaedic surgeons, suggesting that no special training in dysmorphology is required to identify the "CTEV face".

There is a subgroup of children with idiopathic CTEV who can be reliably identified by their facial characteristics. The significance of this finding is unclear but it may be of use in clinical genetic studies, and studies of the aetiology of CTEV.

Keywords : talipes equinovarus ; talipes ; dysmorphology ; facies.

INTRODUCTION

Idiopathic congenital talipes equinovarus is the commonest lower limb malformation, with a reported incidence of 1 per 1000 (*7,25,26*). Several genetic mechanisms have been proposed (*3,15,16,17,19*), suggesting the possibility of genetic heterogeneity (*4*). Environmental factors are also thought to be of aetiological significance (*5,8,9,13,18,20,21,25*).

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In many genetic malformation syndromes, recognisable patterns of facial features can be a useful aid to diagnosis (for example, William's Syndrome (*22*), and assessment of facial dysmorphology is commonly used in clinical genetics. We considered that facial dysmorphology might identify a sub-group of CTEV patients which could be of use in future aetiological studies.

PATIENTS AND METHODS

Ethics approval for this study was obtained from the Multicentre Ethics Committee, ref. WH/MREC/99/0/43, and written informed consent was obtained from the parents (and each child if above the age of 16) of each child included in the study. Parents (and each child if above the age of 16) also consented to publication of the photograph of their children.

Study population

As part of the UK Talipes Study, the parents of 62 children with idiopathic congenital talipes equinovarus (41 males and 21 females) from centres across the UK consented to facial photography. The children had undergone a variety of conservative and operative management regimens. Age ranged from two years and one month, to 16 years and 10 months (mean 5 years 9 months, standard deviation 3 years and 8 months).

Colour digital facial photographs were taken and stored as a *Microsoft® PowerPoint®* presentation. All facial photographs were frontal views.

Photographic analysis

Photographs were reviewed and a list of features seen in some or all of the faces was drawn up (see results).

Five clinical geneticists and three orthopaedic surgeons then viewed all the photographs independently, to assess whether each facial feature was present in each proband. Each feature was recorded as "present" or "absent", giving a range of scores for each feature from zero if none of the team thought the feature was present, to eight if all members of the team thought the feature was present. Each member of the research team documented their assessment independently. Finally, each member of the research team recorded whether they felt that each photograph represented a child with the "CTEV face". All 62 facial photographs were scored using this method.

The photographs were then randomly rearranged, reviewed and scored on a second, separate occasion two weeks after the first viewing to test repeatability of the assessment.

The clinical geneticists on the research team recommended that the following facial features be scored as present or absent in each of the children enrolled in the present study :

1. Frontal bossing
2. Tall forehead
3. Long nose
4. Small jaw
5. Flat upper helix of the ear
6. Prominent ears
7. Epicanthic folds

Statistical analysis

Statistical analysis was carried out using *SPSS© 7.5*. Repeatability was tested using Kappa statistics (*10*), and agreement between the orthopaedic surgeons and the geneticists was tested using Gamma statistics (*11*).

RESULTS

Four examples of children unanimously scored as having the "CTEV face" are shown in figures 1 to 4.

All 62 children were scored for presence of the "CTEV face", but assessment of the individual features in all the photographs was not possible. Hairstyle obscured the forehead in eight probands and the ears in 11 probands. Two photographs were of low technical quality although the features could still be distinguished. The raw data are presented in table I.

Is each feature important in identifying the CTEV face ?

Spearman correlation coefficients for each of the different features and the "CTEV face" were calculated (table II).

A small jaw, frontal bossing, and flat, prominent ears all correlated with the CTEV face with 99% certainty, while a tall forehead and a long nose correlated with the "face" with 95% certainty. Only the presence of epicanthic folds did not significantly correlate with the "CTEV face".

Fig. 1. — Facial photograph of child A unanimously scored as having the CTEV face.

Fig. 3. — Facial photograph of child C unanimously scored as having the CTEV face.

Fig. 2. — Facial photograph of child B unanimously scored as having the CTEV face.

Fig. 4. — Facial photograph of child D unanimously scored as having the CTEV face.

Repeatability

To test repeatability, a kappa correlation was calculated as 0.660 with standard error of 0.080, $p < 0.001$. This demonstrates substantial agreement (*14*) between the two assessments. The ability of the group to identify the CTEV face was therefore highly repeatable.

We also compared scoring for the CTEV face between the geneticists, and the orthopaedic members of the research team using gamma statistics. The gamma correlation was 0.856, with a standard error of 0.055, $p < 0.001$. This demonstrates that there is good agreement between the two groups. The CTEV face is easily recognisable to both geneticists and orthopaedic surgeons.

DISCUSSION

The aetiology of CTEV remains unclear. Identification of syndromic cases may lead to discovery of important aetiological subgroups, and may also have prognostic or therapeutic significance. For example, Alagille *et al* (*1*) described a syndromic form of congenital intrahepatic cholestatic jaundice,

Table I. — Raw data listing number of researchers recording each feature as present for each child (NS : not scored)

FAMILY REFERENCE NUMBER	CTEV FACE PRESENT	INDIVIDUAL FACIAL CHARACTERISTICS						
		frontal bossing	prominent forehead	long nose	small jaw	flat ears	prominent ears	epicanthic folds
20	8	8	6	4	8	1	0	2
22	3	8	6	3	8	7	8	1
34	8	6	7	8	8	6	8	1
37	3	0	8	1	1	0	4	2
40	2	1	8	7	1	NS	NS	3
40	2	4	8	7	1	0	6	5
41	6	0	8	6	6	5	6	2
42	3	NS	NS	3	1	0	0	1
46	1	0	0	7	0	1	3	0
47	2	0	8	1	0	0	1	7
50	2	2	7	0	0	5	4	2
54	0	8	8	0	0	0	0	1
56	0	0	1	3	0	1	1	0
57	0	8	7	3	2	0	0	5
64	0	0	8	0	2	1	1	3
64	8	0	2	7	7	NS	NS	1
66	3	0	8	0	6	NS	NS	0
70	0	0	0	0	0	0	1	0
71	1	NS	NS	8	8	1	0	0
74	3	7	8	0	8	6	7	8
76	4	0	1	6	5	1	0	0
77	6	1	7	0	8	7	3	1
78	4	NS	NS	6	1	NS	NS	0
83	7	5	7	8	2	NS	NS	0
86	3	7	8	4	0	1	1	0
88	7	8	7	2	3	0	2	0
89	3	7	8	8	0	0	7	0
122	0	NS	NS	1	7	NS	NS	0
123	8	8	8	2	3	0	1	0
126	0	1	3	1	0	1	0	0
129	8	7	7	1	8	0	0	8
130	6	4	8	3	8	7	1	0
171	1		3	2	1	1	8	0
173	2	4	7	2	0	6	8	0
511	0	2	0	1	0	0	0	0
583	0	NS	NS	1	0	1	0	0
601	0	1	3	2	0	NS	NS	0
608	5	3	8	7	0	1	0	0
609	7	0	6	1	1	4	6	0
612	4	3	7	8	1	3	7	0

616	1	0	5	7	1	NS	NS	0
620	0	NS	NS	1	1	1	0	0
623	0	0	0	1	0	0	0	0
630	1	0	2	0	0	0	0	0
632	2	2	6	0	0	NS	NS	6
635	5	NS	NS	1	2	2	0	0
636	0	0	7	1	0	0	7	0
675	7	0	6	7	2	NS	NS	1
680	1	1	3	2	1	1	0	0
687	0	0	5	1	0	0	0	0
688	0	NS	NS	1	2	0	0	0
691	2	4	6	1	1	0	0	0
697	6	0	0	1	1	2	8	0
703	3	1	3	0	0	0	0	0
705	7	5	8	0	8	2	2	6
706	0	0	6	2	2	8	5	0
707	2	4	7	1	4	2	1	7
709	4	2	5	0	3	0	1	5
711	3	2	6	2	7	0	7	0
1012	0	2	2	1	1	NS	NS	0
1027	0	1	0	8	1	1	0	0
1033	1	3	8	0	0	0	0	0

identifiable by characteristic facial features, believed to be autosomal dominant, with a better prognosis than other forms of intrahepatic cholestatic jaundice.

Clinical geneticists regularly use facial features in the diagnosis of genetic syndromes and this may lead to elucidation of the genetic defect. For example, Williams *et al* (22), described a syndrome of heart defects, mental retardation and a characteristic face, and 32 years later Curran *et al* (*6*) reported a disruption of the elastin gene as the cause.

We have identified a group of children with CTEV who share similar facial features (the "CTEV face"). A tall forehead with frontal bossing, flat prominent ears, and a small jaw are the most consistent features. Other features may be as significant, but the photographs were all anteroposterior views and in some cases, features were difficult to see. Astley and Clarren (*2*) observed that a key factor for success in this type of work is good quality photographs. They further pointed out that, while some of the individuals in their study looked alike, no two children were identical. Characteristic faces however have the overall "gestalt" (Gestalt, from the high middle German word for shape or form, meaning that a physical entity may be more than the sum of its constituent parts (*24*)). Hall further observed that "a clinical judgement should never be based on a measurable parameter" (*12*).

In our group, although there is a highly significant correlation between the CTEV face and all the individual features, none is absolute. No one feature is seen in all those children identified as having the CTEV face. This is highlighted in figure 5, showing variation in assessment of the presence of the characteristic chin in the seven children unanimously felt to have the CTEV face. The CTEV face is a combination of characteristic features, rather than the presence of all of them.

A number of aetiological theories exist for CTEV, with epidemiological studies suggesting environmental triggers in a genetically susceptible individual. Proposed triggers include intrauterine pressure (*25*), also implicated in the aetiology of

Table II. — Spearman correlation coefficients for all the individual features and the overall characteristic face

	EPICANTHIC FOLDS	FACE	PROMINENT FOREHEAD	SMALL JAW	LONG THIN NOSE	FRONTAL BOSSING	FLAT EARS	PROMINENT EARS
Folds	1							
CTEV face	.233	1						
Forehead	.434**	.342*	1					
Jaw	.270*	.596**	.295*	1				
Nose	-.070	.262*	.020	.215	1			
Bossing	.157	.422**	.517**	.368**	.135	1		
Flat ears	.094	.489**	.275	.493**	.037	.108	1	
Prominent ears	.042	.400**	.243	.227	.204	.141	.654**	1

* correlation significant at 0.05 level
** correlation significant at 0.0 level.

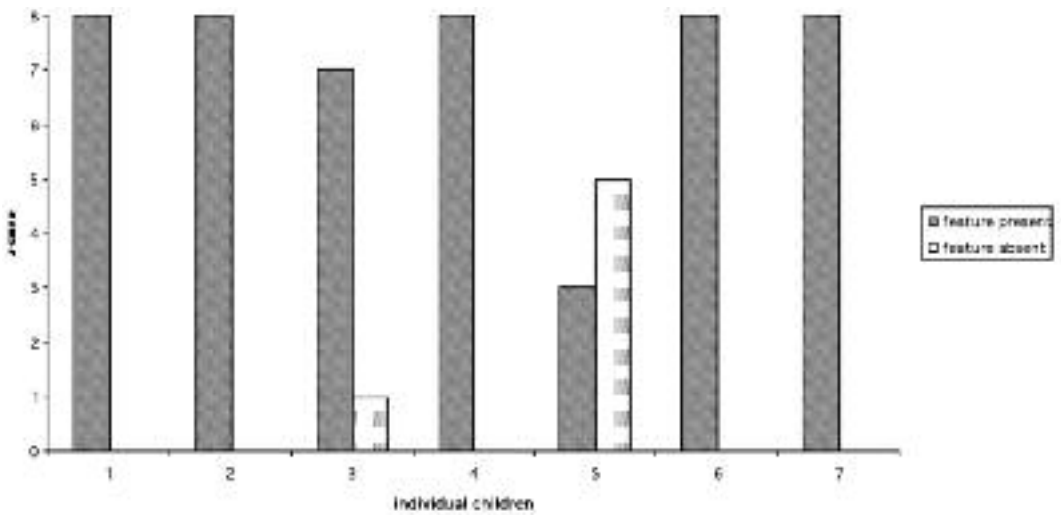

Fig. 5. — Presence of the characteristic chin in children unanimously scored as having the CTEV face

congenital dislocation of the hip (*23*), seasonal viruses (*18*), racial factors (*5,7*), maternal hyperthermia (*9*), viral infections (*20*), and high frequency radio transmitters (*13*). Maternal smoking also appears to be linked to CTEV (*21*).

We have identified a subgroup of children with idiopathic CTEV, identifiable by their facial features, although the significance is unclear. It may represent a subgroup with a particular genetic aetiology, or where particular intrauterine factors have influenced both the development of the face and the limbs.

The present investigation does have some limitations. For example, we only studied Caucasian children within a narrow age range. However, this reflects the typical patient seen in a Paediatric Orthopaedic Clinic in our setting. Nevertheless, our study is based on a relatively large sample of CTEV

patients, with children of different ages. The identification of the characteristic face is highly repeatable, and there is a good correlation between each of the features and the overall face. The scoring system is reliable and repeatable. We hypothesise that the presence of the CTEV face may represent an aetiological subgroup.

CONCLUSIONS

Our study shows that in Caucasian patients there is a subgroup of children with idiopathic CTEV, identifiable by characteristic facial features. These children can easily be identified using good quality photographs taken on reasonable quality cameras without any special equipment or expertise. This information may be of use in studies of clinical genetics and the aetiology of CTEV.

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