Editorial note

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The increasing interest in dysmorphic syndromes is resulting in the identification of numerous cases which do not correspond to any previously delineated disorder. While space generally precludes the lengthy publication of a single patient or family, it is clearly desirable to report such unusual cases. The first of a series of ‘unknown syndrome’ short reports appear here, together with guidelines for preparing such reports. Authors should also generally use this format for case reports of unusual but previously recognised syndromes that are being submitted.

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Guidelines for contributors for unknown syndrome reports and for short reports of diagnosed syndromes

UNKNOWN SYNDROMES
Short case reports of unknown dysmorphic syndromes are welcome; these can be of a sporadic or familial nature. Before submission, the details of the cases should have been presented at a dysmorphology or genetics meeting or assessed by experienced colleagues. Details should be given succinctly. Usually two, but in exceptional cases three, photographs or radiographs can be included. The reports should take the following form.

Summary
List main features, for example, cleft palate, iris coloboma, developmental delay, parental consanguinity.

Medical history
Prenatal and birth history. If not a newborn, list when symptoms and signs occurred.

Clinical examination
Note the age this was performed and measurements and centiles. List findings, starting with general ones such as development, posture, abnormal movements, generalised skin problems, and then describe anomalies by region from head to foot.

Genetics
Family details should be given in all cases, whether positive or negative. In particular, numbers of normal sibs, details of miscarriages, and presence or absence of consanguinity should be noted.

Investigations
Include radiography, karyotype, EEG, EMG, biopsy, necropsy, etc, as indicated.

Discussion
Brief discussion on differential diagnosis (rarely more than one paragraph), possible factors in pathogenesis, and aetiology.

References
No more than five.

DIAGNOSED SYNDROMES
Short reports of diagnosed dysmorphic syndromes are also encouraged, especially for those syndromes where there are few published reports, or where the case(s) reported adds clinical or genetic information. The reports should take a similar form to that for unknown syndromes, discussion should be brief, and references need not be comprehensive but should include any review articles.