Clinical Algorithms

Diagnostic classification of the aetiology of mental retardation in children

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Children with mental retardation are often subjected to a routine battery of tests, a practice that has been criticised.¹ Investigations often include a blood count, a biochemical profile, urinary screening for metabolites including amino acids, and radiography of the skull and wrist for bone age and may be more extensive. Such routine testing of all children with mental retardation is done in the hope of classifying cases in which there are no specific clinical clues to aetiology. Several series have been published, however, in which no unsuspected diagnoses were made in this way.^{2,4} Our aim is to provide a basis of rational investigation planned individually for each patient.

Smith and Simons have suggested primary categorisation according to the time of onset of evident mental retardation.⁴ This, however, leaves 41% of patients unclassified. Our method is based on clinical history and examination and takes the form of a diagnostic sieve.

History and examination

A careful history, with particular emphasis on prenatal, perinatal, and neonatal events, is essential. The mother's obstetric history, including details of any abortions and stillbirths and of the current pregnancy, should be taken. The family history should include full details of ages of parents and siblings and their health and education and a search of the extended family for a history of similarly affected members (which may be found in 10% of cases⁵). A full sociocultural history should be taken and details of development noted. Examination should include an evaluation of the child's general well-being and the plotting of height, weight, and head circumference on appropriate centile charts. Clinical examination should investigate not only associated abnormalities of internal organs but also other congenital abnormalities including minor anomalies of the facies or hands (table).

Three or more associated congenital abnormalities

The finding of three or more congenital abnormalities is significantly more common in children with mental retardation than in normal children or in those with a ventriculoseptal defect or cleft palate.⁵ These children constitute the multiple congenital abnormality and mental retardation group, which included 40% of children with severe mental retardation in the large survey by Kaveggia *et al.*³ This group includes children with chromosomal disorders,

Common anomalies found in children in the multiple congenital abnormality/mental retardation group with idiopathic mental retardation

System	Anomalies
Growth	Primordial short/tall stature
Eye	Inner epicanthic folds, mongoloid slant, mild hypertelorism, strabismus, coloboma, cataract
Ear	Abnormal position, preauricular sinus
Face and mouth	Narrow maxilla (palate), micrognathia, midline sinus
Hand	Abnormal palmar creases, high axial triradii, clinodactyly, syndactyly
Chest and heart	Accessory nipple, pectus excavatum, ventricular septal defect, Fallot's tetralogy
Abdomen and genitalia	Inguinal hernia, cryptorchidism, hypospadias
Skin	Multiple or extensive haemangiomas, café au lait, depigmented patches
Other	Equinovarus deformity, scoliosis, deep sacral dimple

which are now diagnosed with increasing frequency with modern methods of chromosome analysis, and recognised non-chromosomal disorders, the diagnosis of which is facilitated by computer based dysmorphic registers. The identification of a known syndrome may be an indication for genetic counselling.

Primary central nervous system malformation

Children in the primary central nervous system malformation group, which includes those with hydrocephalus, hydrancephaly, encephalocele, spinal dysraphism, and microcephaly, are less often associated with a significant risk of recurrence. There are, however, important exceptions, notably children with spina bifida-hydrocephalus, for whom the risk of recurrence is well recognised, and some with microcephaly that may be inherited as an autosomal recessive trait.

Abbreviations used in algorithm:

CMV=cytomegalovirus	LFT=liver function tests
CNS=central nervous system	MCA=multiple congenital abnormality
CP=cerebral palsy	MR=mental retardation
CT=computed tomography	PKU=phenylketonuria
ECG=electrocardiography	SXR=skull x ray
EEG=electroencephalography	TORCH=serology for toxoplasma, rubella, cytomegalovirus, and herpes
EM=electronmicroscopy	USS=ultrasound scanning
ERG=electroretinography	VER=visually evoked responses
IEM=inborn error of metabolism	
IUGR=intrauterine growth retardation	
IVU=intravenous urography	

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Aetiology of mental retardation in children

Congenital infection

Although congenital infection is responsible for only 2% of mental retardation,³ its variable presentation merits early consideration on the algorithm. If microcephaly, eye problems, or, rarely, the features characteristic of a specific congenital infection are found a careful neonatal history should be taken, with particular regard to intrauterine growth retardation, jaundice, purpura, and hepatosplenomegaly.

Progressive disease

Progressive disease, although uncommon, is important because of prognostic and often genetic implications. Progression may be defined as continuous or intermittent loss of skills, which may be evident from the history when the child is first seen or may become clear with the passage of time. This highlights a fundamental consideration in the use of our algorithm: that the clinician should always be alert to changes in the clinical evidence during follow up, which may mean returning to the beginning of the algorithm and reviewing the classification.

Prominent central nervous system dysfunction

Those with prominent central nervous system dysfunction other than mental retardation include children with cerebral palsy, fits, hypotonia, or noticeably abnormal behaviour who have not been

classified earlier in the scheme. Children in this group are perhaps most commonly subjected to "routine investigations," when there is in fact seldom indication for tests other than electroencephalography in those with seizures.

History of environmental insult

The group with a history of environmental damage is meant to include only those with a clear history of an insult which is temporally associated with the onset of mental retardation and consistent with the findings on examination.

Finally, recent evidence suggests that in 10% of the remaining children with mental retardation in whom no specific diagnosis is made abnormalities of the X chromosome may be found, and this should be excluded.

References

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- 2 Carson NAJ, Neill DW. Metabolic abnormalities detected in a survey of mentally backward individuals in Northern Ireland. *Arch Dis Child* 1962;37:505-13.
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- 4 Smith DW, Simons FER. Rational diagnostic evaluation of the child with mental deficiency. *Am J Dis Child* 1975;129:1285-90.
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(Accepted 9 September 1986)

CLINICAL CURIOS

The case of the missing cards: a clue to the diagnosis of Munchausen's syndrome

A 28 year old woman who was on holiday was admitted with retrosternal chest pain and tachycardia. She had a history of a previous episode of pericarditis and of pulmonary embolism. On examination she was greatly distressed, but the only abnormal findings were mild fever, an ejection systolic murmur, and intermittent sinus tachycardia. The results of investigations showed a slight serial increase in creatinine phosphokinase activity and an increased titre of antibodies to influenza A virus. These abnormalities were not considered to be sufficient to account for her symptoms. In the next two weeks she had several episodes of fever, tachycardia, and considerable distress, but the results of repeated examinations and extensive further investigations were unrewarding. She finally settled and was discharged home but was readmitted later the same day after collapsing with a recurrence of her initial symptoms. Once again the results of examination and investigations were unhelpful.

At this point it was realised that she had been on the ward for two weeks but had received no get well cards or any other mail. There had been no telephone inquiries about her, though she claimed to live with her parents and brothers. A telephone call to her general practitioner (who had not been contacted before because she claimed never to have been ill at home) solved the problem. She was known to have Munchausen's syndrome and had been admitted to at least 70 different hospitals, with a personal best of four admissions to separate hospitals on the same day.

The "missing cards" sign does not seem to have been described before, though Shah *et al* suggested that in their patient with Munchausen's syndrome the total absence of visitors should have alerted them to the diagnosis.¹ For the hospital patient greetings cards and visitors may be the only manifestations of normal—or abnormal—social relationships outside hospital.

In a second case get well cards again gave us an insight into the clinical problem. A 38 year old diabetic woman had had a series of admissions for ketoacidosis. Once on the ward she would greet the staff by their first names and was happy to help with ward tasks. We suspected that some of her admissions might have been self induced, and this suspicion was strengthened by finding that her bedside locker "blossomed" with a fine display of get well cards within a day of each admission. Closer inspection showed that the cards appearing after each admission were identical and that several had been signed by the same person. We believe that this patient, who was lonely

and may have had a personality disorder, if not classic Munchausen's syndrome, hoarded these cards and brought them in with her whenever she was readmitted.—DAVID EVAN MORRIS, medical registrar, Glan Clwyd Hospital, Bodelwyddan, Clwyd.

- 1 Shah KA, Forman MD, Friedman HS. Munchausen's syndrome and cardiac catheterisation. *JAMA* 1982;248:3008-9.

Might the regular use of a visual display unit cause headaches? The patient sat about 12 inches away from the side of a unit connected to an autobank throughout the day for three years.

Many studies of visual display unit operators have reported complaints of headache. These have usually been attributed to ergonomic factors such as eyestrain and poor posture.¹ It is difficult, however, to apply such considerations to the correspondent's patient who sat beside the visual display unit and presumably did not operate it. Non-ionising electromagnetic radiation may be emitted from visual display units. This radiation has an extremely low frequency component (50-60 Hz) analogous to that found beneath overhead power lines. It has been suggested that exposure to such fields may be associated with symptoms, including headache. The few measurements made on visual display units have, however, shown electric and magnetic field strengths of only 1-10 V/m (rms) and 0-22-0-56 A/m (rms) respectively. This is not much different from ambient levels in laboratories and homes, and much weaker than those around domestic appliances.² Electromagnetic radiation may also have an inaudible acoustic component. This may be sonic (15-20 kHz) or ultrasonic (20-32 kHz) and levels up to 68 dB have been measured. Some individuals may be sensitive to this and headaches have been reported.³ After servicing of the visual display units the "sound"—and the headaches—disappeared.—W R LEE, professor, and A R SCOTT, lecturer, in occupational health, Manchester.

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- 2 Marriott IA, Stuchley MA. Health aspect of work with visual display terminals. *J Occup Med* 1986;28:842.
- 3 Chatterjee DS, Crookes AJ. VDUs and relative annoyance from high frequency noise. *J Occup Med* 1984;26:552-3.