

## *Personal practice*

# Pitfalls in developmental diagnosis

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**SUMMARY** I have never seen a paper or chapter of a book devoted to pitfalls and mistakes in developmental diagnosis. This paper is designed to try to fill the gap. It concerns the avoidance of mistakes in developmental diagnosis and is based entirely on mistakes that I have made myself and now learnt to try to avoid and on mistakes that I have seen, most of them repeatedly. I have made no mention of mistakes that could theoretically be made but that I have not personally seen.

I believe that most assessment errors are due to overconfidence and to the view that developmental diagnosis is easy. Many other mistakes are due to reliance on purely objective tests with consequent omission of a detailed history and physical examination, so that factors that profoundly affect development but are not directly related to the child's mental endowment are not weighed up before an opinion is reached.

It is a mistake to think that developmental diagnosis is easy and requires little training. It often is easy, but it may be extremely difficult. Errors cause much parental anxiety and may be tragic for the child.

Failure to recognise a normal variation in some aspect of development may lead to unnecessary investigation or treatment, worry for the parents, and disadvantage for the child. The common cause of erroneous dogmatic statements about development is overconfidence.

I know of no article on pitfalls in developmental diagnosis apart from a brief discussion in the third edition of Gesell and Amatruda's 'Developmental diagnosis'.<sup>1</sup>

### **General principles**

I believe that certain general principles must be observed if pitfalls in developmental diagnosis are to be avoided. They are as follows.

(1) An essential requirement is a thorough knowledge of the normal (better termed the average) and the normal variations that do not amount to disease and an effort to try to understand the factors that cause these variations. A thorough knowledge of the normal is a necessary preliminary to the study of disease.

(2) Children differ widely in all aspects of development—so widely, that it is impossible, de-

spite the claims of some workers,<sup>2</sup> to define the range of normal. It would be convenient if the range of normality could be delineated, but it is impossible. A line can never be drawn between normal and abnormal. All that can be said is that the further away from the average a child is in any field the less likely he is to be 'normal'. Failure to recognise this leads many to declare that a child 'should' reach milestones, such as sitting unsupported, walking without help, talking, or controlling the bladder, by a specified age, as if all normal children are the same, with no normal variations. Such views, relayed to parents after developmental assessment, may cause great anxiety. I have heard of parents being told that their 6 month old baby has 'failed' his test and of 8 month old babies being referred to psychiatrists because of suspected lateness in passing certain milestones.

(3) A child's level of development is the end result of a wide variety of factors—prenatal, perinatal, and postnatal. Many of these are not directly related to the child's mental endowment yet have a profound effect on his development. Failure to take these factors into consideration helps to explain the poor correlation so often reported between psychological tests in infancy and later intelligence quotient (IQ) scores, the assessment having been made solely on the basis of objective tests.

(4) The mentally subnormal child, unless the

subnormality is of later postnatal origin, is retarded in all aspects of development, except sometimes in gross motor skills, and rarely in sphincter control. Hence we can never diagnose mental subnormality on backwardness in just one or two fields of development.

(5) Some fields of development are far more important than others for developmental assessment. The least important in infancy is the most easily scored—namely, gross motor development (sitting and walking). Some mentally subnormal infants show average motor development, while advanced motor development certainly does not signify a high level of intelligence.<sup>3</sup> The most important features for the assessment are features that cannot readily be scored because norms have not been established, features that Gesell termed 'insurance factors'—the child's alertness, responsiveness, interest in surroundings, persistence, concentration, and the glint in the eyes. Other features are his memory, understanding, and the quality of his vocalisations (which is difficult to assess without special equipment).

Manipulative development is far more important than gross motor development. Important developmental features, rarely seen in case notes, include ambidextrous (average 20 weeks) or unidextrous (average 28 weeks) approach to an object, the rapidity with which he accidentally drops a cube, the question of whether he drops one cube when offered another, tremor or ataxia or other abnormal hand movement when reaching for an object, the transfer of a cube from one hand to another (normally 6 months), and the quality of the grasp (evolving from the crude palmar grasp of the cube at about 5 months to the mature grasp between the tip of the thumb and the tip of the forefinger at 11 to 12 months).

The index finger approach to an object is an important and easily observed feature from 9 to 10 months of age. If I see the index finger approach to an object I would think that the child's *overall* mental development could not be less than that of a 9 to 10 month baby. The 'pincer grasp', by which one means the grasp of a pellet or small object, refers only to the grasp between the tip of the thumb and tip of the forefinger; it develops at the same time as the index finger approach: so does the 'matching' of cubes—the child apparently comparing the cube in one hand with that in the other. When he holds out a cube to his mother, it is noted whether he withdraws it (average 44 weeks), refusing to let it go, or shows the later skills of releasing it into her hand (average 48 weeks). It is not enough to record just *whether* a child has acquired a certain skill: one has to note the maturity and the rapidity

with which he does it. For instance, in the crude screening test for hearing, it is noted not only *whether* he turns his head to sound, but the *rapidity* of the response, and his later ability to turn to sound made below the ear, or later still above the level of the ear.

It is thoroughly misleading to obtain a score for different aspects of development, as in the Griffiths tests,<sup>4</sup> obtain the average, and then regard that as the 'IQ'. Failure to recognise the different developmental importance of various aspects of development again helps to explain the relatively small correlation between psychologists' tests in infancy and subsequent IQ scores.

(6) A clinical developmental diagnosis should always be based on the history, full physical and developmental examination, special investigations where relevant, and the interpretation of the results. The diagnosis must be made on the child as a whole. It is a mistake to make it purely on the basis of examination of one little bit of the child by a few objective tests in a limited field of development: such an approach inevitably omits vitally important facts. Neither should it be made as a 'spot' diagnosis. It is usually easy at a glance from a distance to diagnose Down's syndrome and some other forms of mental subnormality, but it is difficult for parents to accept a doctor's 'spot' diagnosis, followed by his confident statement on the outlook.

It is easy to be misled by a child's ugliness, odd facies (often taking after one of his parents), charm, facile conversation (in the case of hydrocephalus), difficult behaviour, or isolated advancement or retardation in one field.

### The history

To avoid mistakes by the omission of relevant, highly important factors that affect development, a detailed history is essential. It will include all prenatal, perinatal, and postnatal factors. These will cover the main 'risk' factors in pregnancy, such as infections, placental insufficiency, hypertension, antepartum haemorrhage, adverse socioeconomic factors, malnutrition, genetic and familial factors, and drug taking, including drugs of addiction. It will include in particular factors associated with the aetiology of mental subnormality and cerebral palsy. The importance of these risk factors must not be exaggerated—a matter of importance when a child's suitability for adoption is being considered. When there is doubt about his level of development the history of a risk factor, such as mental subnormality in a parent, serves only to increase that doubt. But a risk factor itself should never lead to a hasty diagnosis that a child is mentally subnormal. A

mentally subnormal parent can have a normal or mentally superior baby.<sup>3</sup>

The family pattern of development is important, particularly with regard to backwardness in motor skills, speech, sphincter control, or learning disorders or to backwardness in early weeks followed by normal or advanced development.

If serious mistakes are to be avoided appropriate allowance for the history of preterm delivery must be made. If the baby was of low birth weight the duration of gestation must be known to distinguish intrauterine growth retardation. The Griffiths and other developmental tests do not make such an allowance. Whereas an average full term baby begins to smile in response to his mother's overtures at 4 to 6 weeks, the baby born three months early would be expected to smile at 4 to 6 weeks plus 3 months. I have seen babies born two or three months early assessed for adoption 6 weeks after birth.

Few would fail to obtain a history of perinatal 'risk' factors, but these factors are often misinterpreted.<sup>5</sup> It is a widespread practice to ascribe mental subnormality or cerebral palsy to 'brain damage' or 'birth injury' due to hypoxia or breech or forceps delivery, without considering the possible or likely prenatal causes of those conditions. I recently reviewed this subject, pointing out that cerebral palsy commonly occurs after normal labour, with no perinatal problems, while severe delivery problems, including pronounced hypoxia, are usually not followed by evidence of 'brain damage'.<sup>6</sup> Failure to recognise prenatal factors as the main cause of so called 'brain damage' has major medicolegal consequences and results in errors of counselling about the risk of future pregnancies.

The history must include postnatal factors that may profoundly affect development and that are unrelated to his mental endowment. For instance, defective weight bearing in an otherwise normal baby is usually due merely to his not being allowed to bear weight on the legs, for fear that he will develop rickets, bow legs, or knock knee. Yet some tests—for example, Ruth Griffiths—ignore this and give a low score as a result. Other aspects of parental management, stimulation or lack of it, socioeconomic problems, emotional deprivation, and the chance given the child to learn to feed himself, dress himself, and attend to his toilet needs need to be known. It is important to know if the mother is out at work all day. The paediatrician must know about malnutrition, illness, head injury, exposure to toxic substances, symptoms suggestive of cerebral palsy—for example, the hand or hands being kept tightly closed at an age when the hands

should be largely open or asymmetry of limb movements as in hemiplegia.

Precision in history taking is necessary if errors are to be avoided. It is essential that parent, doctor, or nurse should understand exactly what the other means. The doctor, on the basis of his experience, has to decide how reliable is the history and how much of it he is able to accept. It is a mistake not to ask the mother about her child's development: the reliability of her story must be checked against the doctor's own objective findings. It is not enough to ask whether the child smiles, rolls over, grasps objects, sits, creeps, walks, talks, or is 'dry'. It is not enough merely to know *whether* a child does a thing, but *when* he began to do it, and how often, and with what degree of maturity.

Developmental histories that I experience are commonly of very limited value because they are imprecise. The milestone 'beginning to smile' refers only to smiling in response to the mother's overtures and not to a grimace or twitch when she tickles the face or to the baby's facial movement in sleep. 'Rolling over' refers to rolling *completely* over, and rolling from prone to supine has to be distinguished from rolling from supine to prone, which usually comes later. 'Grasping objects' means going for an object without it being put into the hand and has to be distinguished from the grasp reflex, or the ability to hold an object placed in the hand. 'Sitting' means sitting on the floor or another hard surface for seconds, either with the hands forward for support or without support. It does not mean sitting with support in the pram. 'Creeping' (on hands and knees) has to be distinguished from the earlier 'crawling' on the abdomen. 'Chewing' has to be distinguished from 'sucking'—for example, a biscuit. 'Walking' means walking a few steps without support; 'talking' consists of saying words with meaning, not just 'mumum, dadada'. 'Toilet trained' means that the child is mainly dry and clean day and night and has to be distinguished from the much earlier conditioning when the child voids as the buttocks feel the rim of the potty, whether awake or asleep. If control is incomplete the stage reached should be recorded—for example, the child telling the mother that he has voided, or, later, that he is about to void.

I commonly see in notes 'held her head up' at x weeks or 'bears weight on the legs'. These words are meaningless without definition. Newborn full term babies can hold the head up momentarily or bear some weight on the legs. Other information needed concerns the age at which the child began to play games and to have skills involving imitation and memory—for example, peep-bo, clap hands, waving 'bye-bye', helping to dress by holding the arm out

for a sleeve or foot out for a shoe, and feeding himself by picking up an ordinary cup, drinking from it, and putting it down without help and without much spilling.

A properly taken history includes the *rate* of development, indicating that the rate is steady, improving, or slowing, vital information necessary for assessing a difficult case.

### **The physical examination**

For developmental assessment, a full physical examination is essential if serious errors are to be avoided. This will include examination for neurological and physical handicap, congenital anomalies, and defects of vision and hearing—all conditions that may profoundly affect development and developmental tests but that may have no direct relation to the child's level of intelligence. Due allowance for these handicaps must be made, for otherwise his real ability and potential may be seriously underestimated. A physical disability that is easily missed in a child who is later than usual in gross motor skills is Duchenne muscular dystrophy: the diagnosis is complicated by the lower than average level of mental development commonly found.

An essential part of the physical examination is the head circumference in relation to the child's weight. Serial measurements are essential if there is doubt, for they may show a falling off from the centile distribution, which is a most important indication of brain defect. Other causes of unusual head measurements, such as familial traits, must be known. I myself made a mistake in measurement by using a tape measure that had become inaccurate by stretching.

Irrelevant physical features include the age of closure of the anterior fontanelle, an epicanthus, or central palmar crease; the latter may be found in normal children.

During the physical examination general features are noted, such as unusual behaviour (as in autism), the quality of vocalisation, the child's interest in surroundings, and responsiveness, and signs suggesting mental subnormality, such as bruxism when awake, persistence of 'hand regard' after 20 weeks, or mouthing, slobbering, and casting at an age when he should have grown out of it.

### **The developmental examination**

Whatever the developmental test, it must be ensured that the child's performance seems to be the best of which he is capable. He should not be tested when he is on drugs or is tired, sleepy, poorly,

hungry, or bored. When he does badly in a test a decision has to be made about whether he was really trying. The older child may refuse to take part in a test because he is shy or regards it as too easy or just silly.

I think that it is a mistake to separate the infant or small child from his mother. It adds to the difficulty of the child not achieving his best in strange surroundings. The mother has to resist the urge to help the child in the tests. Rigid adherence to a method of testing is undesirable: the tester should adjust the order of tests as soon as he sees that the child is not interested or is becoming bored. One never corrects a child or says 'no' when he performs a test incorrectly.

In a busy obstetrics hospital follow up clinic or a child health clinic tests must be practical. I know a book that describes 115 different methods of testing, occupying up to 125 minutes for each child. Other popular tests take 40 minutes or more.

I feel that some do not sufficiently distinguish that which is interesting—for example, many of the 70 or more primitive reflexes—from that which is important. As far as I can determine, the only primitive reflexes that can help in diagnosis are the grasp reflex, Moro reflex, and asymmetrical tonic neck reflex. Some would add the oral and parachute reflex. It is useful to ask how a particular test would help the diagnosis and what condition would be missed by not doing it.

Once a test has been chosen—for example, as described by Gesell or anyone else—the materials used must be the same as those on which the norms were established. I often see children tested with cubes of varying sizes, whereas Gesell's norms were based on one inch cubes.

### **Interpretation**

It is said that intelligence tests are more a test of the intelligence of the examiner than the examinee. The examiner must allow for all factors, including physical and sensory handicap, which greatly affect development, especially when they have little to do with the child's mental endowment. Cultural and ethnic factors may considerably affect a test score, but there is no satisfactory way of allowing for them except by judicious guesswork.

I often see incorrect interpretation of a test because extenuating factors have been missed. For instance, a 3 or 4 year old's poor performance on repetition of digits may be due to a hearing defect or failure to understand what he is asked to do. A poor performance on motor or other tests may be due to hypotonia, hypertonia, a visual defect, or mere lack of opportunity to practice—for example, weight bearing.

A 6 or 8 week infant placed in the prone position will revert to the fetal position if asleep and may be given a low score as a result. Advanced development in the prone position may be due to cultural factors,<sup>7</sup> the child having been encouraged to lie and sleep in that position.<sup>8</sup> Other causes of delayed motor development are often missed—familial traits, personality (fear of falls), shuffling, or Duchenne muscular dystrophy. It is usually a mistake to ascribe late motor development to obesity or a dislocated hip. Delayed sitting may be due to congenital shortening of the glutei, gastrocnemius, and hamstrings.<sup>9</sup> A poor performance in drawing tests, or in identification of pictures, may be due to the child not having been given a chance to learn. Many ascribe delayed toilet training purely to psychological problems, ignoring the familial factor, delayed maturation of the relevant part of the nervous system, the sensitive period for learning, or urinary tract infection or anomalies. Psychological problems may well be superimposed on other factors, without being the primary cause.

An important source of error is the popularity of a unitary score in developmental assessment and misinterpretation of its accuracy and predictive value. Knobloch and Pasamanick have rightly deprecated this.<sup>1</sup> They discussed the confusion concerning the terms 'developmental quotient' (DQ) and 'intelligence quotient' (IQ). The DQ indicates how far a baby has progressed in *all* aspects of development, especially behaviour, in relation to the average for his age. The IQ relates his age to his performance, mainly in verbal and problem solving tests, on the basis of pass or failure. The DQ is profoundly affected by the environment and many other factors unrelated to his genetic endowment.

No one has satisfactorily defined 'intelligence', but many would agree that it cannot be described as a single score or figure, for there are many different types of intelligence and of human abilities.

It is unwise, especially in the young infant, to give an exact figure for a test score. I was asked to see an older child with a learning disorder: he had been seen by two psychologists, who independently reported an IQ of 79 and 79.1. An 'exhibit' in a clinical meeting showed that a 6 week old baby with a metabolic defect had an 'IQ' of 70 on the Griffiths scale and at 12 weeks had an 'IQ' of 76—conclusive proof of the efficacy of treatment! Such accuracy is impossible. One reason is the fact that the so called 'norms' of development, on which most of the commonly used developmental tests are based, were established on a highly selected population, instead of on the population as a whole. I briefly reviewed this matter with reference to the developmental tests described by Gesell, Beintema, Brazelton, and the

Denver and Kansas Groups.<sup>10</sup> The tests were based only on children of white origin who were full term and mostly had no prenatal or perinatal risk factors, such as breech delivery, socioeconomic problems, or those thought to have any developmental abnormality. It follows that we have no valid norms with which to compare the development of children not satisfying these criteria and accuracy is impossible.

When, as often happens, there is much scatter in a child's development in different fields I pay particular attention to Gesell's 'insurance factors'—the alertness, responsiveness, and interest that the child shows—even though he is backward in many or even all fields of development. I believe that I am less likely to err if I pay more attention to his positive achievements, those skills (other than gross motor development) in which he does best, than to the negative ones in which he is most retarded.

In making his assessment of the child's potential the doctor has to try to decide whether backwardness in various fields has been due to adverse environmental factors, such as illness or emotional deprivation, how much is related to physical handicap, and how much is potentially reversible.

There are important pitfalls to avoid in the interpretation of signs that may sometimes indicate cerebral palsy, mental subnormality, and visual or hearing defects. Cerebral palsy cannot be diagnosed on the basis of isolated retardation in motor development nor on the basis of exaggerated tendon jerks (in the young baby) or even persistent ankle clonus, for those signs may disappear as the baby gets older. There is constant confusion about the plantar responses in the first year or so: they are flexor unless there is disease of the pyramidal tracts. Excessive extensor tone is readily missed, and I have made the mistake myself. It is a highly important pointer to cerebral palsy. The infant seems to have good head control in ventral suspension and in the prone position but gross head lag when pulled up from the supine to the sitting position. Resistance is felt when pulling him up to the sitting position because of spasm of the erector spinae, glutei, and hamstrings; the knees flex, so that he cannot sit with the legs extended; he may tend to rise on to his legs, wrongly suggesting good motor development; and each time he is seated leaning forward he falls backwards.

I have made the mistake of diagnosing adductor spasm, on account of limited abduction at the hip, when the limited abduction was due to muscle contracture due to the hypotonic child constantly lying in one position, and I diagnosed the spastic form of cerebral palsy on account of limited joint extension, when the correct diagnosis was punctate epiphyseal dysplasia.

Minimal involvement of the upper limbs, as seen when the child is going for cubes or building a tower, is easily missed, so that spastic paraplegia is diagnosed when the correct diagnosis is spastic diplegia. Spastic paraplegia should alert the doctor to the possibility of a spinal lesion rather than a cerebral one. Spastic monoplegia is extremely rare: I saw only one possible case in my personally observed series of over 750 cases of cerebral palsy.

Toe walking is often thought to be due to cerebral palsy when it is only a normal variant or is due to rarer causes, such as congenital shortening of the Achilles tendon, Duchenne muscular dystrophy, unilateral hip dislocation, autism, or dystonia musculorum deformans. Spastic children are often wrongly thought to have a 'mixed' form of cerebral palsy because the characteristic awkwardness and splaying out of the hands is thought to be athetosis.

I have often seen a late walker thought to have the ataxic form of cerebral palsy because shortly after first walking without support he is unsteady on his feet—like the younger child when he walked alone at a much earlier age. Delayed walking in a mentally normal child is commonly no more than a familial feature: it is most unlikely to be due to congenital dislocation of the hip and almost certainly not due to obesity.

I have often seen infants thought to be blind, because of delayed eye following, when the delay was due only to mental subnormality: in the same way I have seen mentally subnormal children thought to be deaf, because of delayed response to sound. Delayed visual maturation<sup>11</sup> is a rare cause of apparent blindness: but the absence of nystagmus or abnormal ophthalmoscopic findings in the presence of otherwise normal development would avoid the mistake of diagnosing mental subnormality.

Isolated delay in speech is never due to mental subnormality. In a mentally normal child the commonest cause is a familial trait; deafness is a less common cause. It is not due to tongue tie, jealousy, or 'everything being done for him'. Every child learns the meaning of words long before he can articulate them, and some have learnt the meaning

of scores of words without being able to articulate any. The common lulls in the development of speech when a child is learning to walk are a common cause of anxiety.

### Conclusion

After a full history, physical and developmental examination, and interpretation of the findings it is possible to say much about a child's developmental potential and something about his talents but not to say what he will do with them. That will depend on many factors in the future—the quality of his home, friends, and school, his personality, health, and nutrition, and the opportunities that he will have.

Developmental assessment can be very difficult. The wise doctor will learn from mistakes, and to avoid pitfalls in the future, he will try his best to follow up all children presenting unusual developmental features—and there are many of them.

### References

- <sup>1</sup> Knobloch H, Pasamanick B. *Gesell and Amatruda's developmental diagnosis*. 3rd ed. Maryland: Harper and Row, 1974.
- <sup>2</sup> Frankenburg WK, Dodds JB. The Denver developmental screening test. *J Pediatr* 1967;**71**:181–91.
- <sup>3</sup> Illingworth RS. *The development of the infant and young child, normal and abnormal*. 9th ed. London: Churchill Livingstone, 1987.
- <sup>4</sup> Griffiths R. *The abilities of babies*. London: London University Press, 1954.
- <sup>5</sup> Task force on joint assessment of prenatal and perinatal factors associated with brain disorders. *Pediatrics* 1985;**65**:457–8.
- <sup>6</sup> Illingworth RS. A paediatrician asks—why is it called birth injury? *Br J Obstet Gynaecol* 1985;**92**:122–30.
- <sup>7</sup> Super CM. Environmental effects on motor development. The case of African infant precocity. *Dev Med Child Neurol* 1976;**18**:561–7.
- <sup>8</sup> Holt KS. Early motor development. Postural induced variations. *J Pediatr* 1960;**57**:571–5.
- <sup>9</sup> Illingworth RS. Norms for development. *Dev Med Child Neurol* 1986;**28**:122–3.
- <sup>10</sup> Cole GF, Hungerford J, Jones RB. Delayed visual maturation. *Arch Dis Child* 1984;**59**:107–10.

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