THE DEVELOPMENT OF A CHILD WITH POTTER’S SEQUENCE DUE TO UNILATERAL RENAL AGENESIS

Carmen Torres, José Buceta and Josefa Lorenzo

Introduction

Potter’s sequence comprises the consequences arising when kidney development in the foetus is absent or defective. It includes pulmonary hypoplasia, characteristic facial traits (flat, low-set ears, prominent epicanthic folds, increased eye-to-eye distance, flat nasal bridge and receding chin) and other growth anomalies (defective limbs, highly arched feet and broad, clumsy hands with short fingers and - frequently - excess dorsal skin) (Kaffe et al., 1977). About 60% of patients have a bell-shaped chest (Leonidas et al., 1982), which together with pneumomediastinum/pneumothorax (air in the mediastinal and pleural cavities) is a diagnostic sign. Other common abnormalities include retractile testicles, uterine or hemi-uterine agenesis, and cardiac anomalies. Cases featuring hydrocephalus and involvement of the cerebral cortex have also been reported. Most cases due to renal agenesis are spontaneous, although cases of autosomal recessive, autosomal dominant and polygenic inheritance have also been recorded (Kaffe et al., 1977).

The gestation of children exhibiting Potter’s sequence is characterised by oligohydramnios (amniotic fluid deficit), small size for any given foetal age, and premature birth. The oligohydramnios, which is almost always indicative of anomaly, may be caused by a deficit of foetal urine due to the renal anomalies (Thomas and Smith, 1974); in turn it probably causes foetal compression and hence the anomalies in limb position and facial traits. Prognosis depends on the severity of the anomalies, but is generally
poor, and references to the post-natal
development of children with Potter’s se-
quence are correspondingly few. In this
work we describe the development of a
child with a diagnosis of poly-
malformation syndrome with facies Pot-
ter who since the age of 23 months has
been given stimulation treatment at home
and in our Early Intervention Unit.

Subject and Methods

Subject

The subject was the male second child
of a healthy 32 year old mother with no
relevant family medical history. Gestation
and delivery (with cephalic presentation)
were supervised. At birth the child
weighed 2,800g and had a rating of 9-9 on
the standard Apgar test of neonatal well-
being (Apgar, 1953). He remained in hos-
pital for 5 weeks, and was re-admitted at
the age of 4 months and at the age of 6
months, when he was operated on for in-
guinal hernia. From birth, feeding was
hampered by the absence of the sucking
reflex, and was affected via a nasogastric
tube. In medical examinations carried out
during the first few months the subject
was conscious and receptive, and exhib-
ited a good general condition, including
good active and passive mobility, but was
microsomic, disharmonic, pale-skinned
and slightly hypotonic, had scant though
normally distributed subcutaneous fat,
and exhibited psychomotor retardation,
Potter facies (large low-set ears with
broad helices, epicanthic folds, ocular
hypertelorism, narrowed palpebral fis-
sures (especially the left), broad, shallow
nasal bridge, microretrognathia, cranio-
facial disproportion, short neck, arched palate and dental malposition) and other
non-facial Potter sequence features:
thoracic asymmetry with a prominent
right hemithorax, pectus excavatum and
widely separated nipples; small scrotum
(right testicle retractile, left high); and
ciliodactylism of the fifth fingers, right
hand with a single palmar crease, anoma-
lous implantation of the second and
fourth toes of both feet, and pedal valgus.

The subject’s karyotype was 46xy. Suc-
cessive ultrasonograms showed no left
kidney and enlargement of the right,
with frequent episodes of hydronephro-
sis. Cerebral ultrasonography showed
moderately dilated ventricles. An electro-
encephalogram, recorded at the age of 15
months, indicated intense diffuse altera-
tion of cortical and subcortical structures,
especially in the temporal regions. Mag-
netic imaging performed in view of a
gradual increase in cranial circumference
showed alterations of white matter sug-
gestive of encephalopathy, but no signs of
hydrocephalus.

At the age of 12 months the subject be-
gan to attend physiotherapy sessions
three times a week. At the age of 18
months he was able to maintain his head
upright, at 21 months he began to walk,
and at 22.5 months he began to attend
stimulation session (see below, Proce-
dure). Both stimulation and physi-
otherapy have continued to the present
day. At the age of 2 years he began to use a
rigid corset to reduce the thoracic defor-
mation and assist upright stance. At the
age of 2.5 years swallowing was normal
and the nasogastric tube was withdrawn.

Instruments

The Brunet-Lezine Scale for Measuring
Psychomotor Development in Early
Infancy (Brunet and Lezine, 1965) quanti-
fies development in four areas (posture
control and motoricity, hand-eye
coordination and relationships to objects, language and sociability) as both a developmental quotient score and a developmental age (DA).

The Battelle Developmental Inventory (Spanish version by De la Cruz and Gonzalez, 1996) comprises a total of 341 items grouped into the areas corresponding to personal-social adaptive, motor, communication and cognitive skills. Scores are expressed as percentiles, equivalent age (EA), and z scores (scores normalised to have a mean at zero and a standard deviation of unity in the reference group used to develop the test). In this paper we report equivalent ages and z scores.

Procedure

At the age 22 months 17 days the subject entered a stimulation programme run at the Early Intervention Unit of the University of Santiago de Compostela by a team of psychologists and teachers specialised in early intervention. On the basis of the Brunet-Lezine Scale results, an initial personalised protocol was designed by adapting the objectives and activities listed in The Portage Guide to Early Education (Bluma et al., 1972) in view of the subject's characteristics and deficits in each of the five areas considered by the Guide: socialisation, language, self-help, cognition and motoricity. Therapy was applied in the Unit in three one-hour sessions each week, and a copy of the protocol was given to the parents, who were individually instructed concerning its application at home and the auxiliary material that they might use. We emphasised both the subject's need for daily stimulation and the desirability of a flexible approach exploiting everyday interactions for application of the protocol. When the objectives set in the first protocol had been achieved, a second was designed and applied, and so on. Each protocol was designed for use over roughly a three-month period, although the time for which it was actually employed naturally depended on the subject's progress. By way of example, the protocol introduced at the age of 35 months is shown in the Appendix.

Since the boy's entry into the stimulation programme, his physical development and social and family relationships have been monitored by collaborating paediatricians and social workers. He has been re-evaluated on the Brunet-Lezine Scale or the Battelle Developmental Inventory approximately every 6 months. Here we report Brunet-Lezine results obtained at entry, and at the ages of 28 months 11 days and 36 months 17 days, and Battelle Developmental Inventory results obtained at the ages of 53, 59 and 65 months.

Results and Conclusions

The Brunet-Lezine Scale results obtained at the ages of 22, 28 and 36 months (TABLE I) show the subject's positive progress during this period in all the developmental areas considered by the scale. Between the chronological ages of 22 and 36 months, the overall developmental age increased from 13 to 27 months. Improvement was greatest in the area of posture control and motoricity, in which DA rose from 10 to 27 months, and in the area of language, in which a rise from 9 to 26 months occurred. In the areas of coordination and socialisation, DA increased by 13 months, i.e. just slightly more than the chronological age.

65
TABLE I
Brunet-Lezine Scale Developmental Age (in months) at chronological ages 22, 28 and 36 months

<table>
<thead>
<tr>
<th>Chronological age (months)</th>
<th>P</th>
<th>C</th>
<th>L</th>
<th>S</th>
<th>Total Scale*</th>
</tr>
</thead>
<tbody>
<tr>
<td>22</td>
<td>10</td>
<td>17</td>
<td>9</td>
<td>12</td>
<td>13.3</td>
</tr>
<tr>
<td>28</td>
<td>13.5</td>
<td>24.1</td>
<td>18</td>
<td>15.5</td>
<td>19</td>
</tr>
<tr>
<td>36</td>
<td>27</td>
<td>30</td>
<td>26</td>
<td>25</td>
<td>27</td>
</tr>
</tbody>
</table>

P - posture control and motoricity; C - hand-eye coordination and relationship to objects
L - language; S - sociability
* Note that the Total scale DA is not necessarily the mean of the subscale DAs, see Brunet and Lezine (1965)

TABLE II lists the z scores and equivalent age calculated from the Battelle Developmental Inventory results recorded at the chronological ages of 53, 59 and 65 months. In the area of personal-social skills, EA rose during this 12 months period from 44 to 55 months, i.e. less than the chronological age. Although scores for interaction with adults, coping and social role were all quite close to the mean for the general population at the age of 65 months, scores for self-concept, interaction with peers and expression of feelings/affects were still low; in fact, all these latter three subareas showed regression from rather better scores obtained 6 months previously.

In the area of adaptive skills, the EA rose from 45 to 58 months. Scores for dressing, toileting, personal responsibility and capacity for attention all approached or exceeded the mean for the age group, particularly marked progress having been made in dressing and responsibility. The only subarea still showing marked deficiency at the chronological age of 65 months was eating, which still required aid.

In the motor area there was a significant contrast between coarse and fine motor control. Coarse motor control was still significantly deficient at the age of 65 months in spite of the spectacular advance in EA from 16 to 39-42 months during the previous year; deficits in body coordination and locomotion may have been partly due to the subject’s macrocephaly and corset. By contrast, fine muscle and perceptual motor control both improved to near-average levels, with the result that the overall EA for fine motor control rose by 19 months to equal chronological age at 65 months.

The subject’s overall EAs for communication and cognition at the chronological age of 65 months were both 69 months, having risen by 18 and 16 months respectively during the previous year. Expressive communication skills, which at the age of 53 months had lagged somewhat, had overtaken receptive communication skills by the age of 65 months; in particular, the subject had practically mastered the use of frontal phonemes and consonant groups in spontaneous speech. At the age of 65 months his language was fluid and perfectly comprehensible in spite of persistent (though improved) generalised hypotonia, which particularly affected the orofacial region. Respiratory insufficiency gave his voice an unusual quality, but did not diminish comprehensibility. The rise in cognitive EA affected almost all cognitive areas, quite satisfactory scores being achieved for perceptual
### TABLE II
Battelle Developmental Inventory z scores and equivalent ages (EA) at chronological ages 53, 59 and 65

<table>
<thead>
<tr>
<th>Area/Subarea</th>
<th>Chronological Age in months</th>
<th>53</th>
<th>59</th>
<th>65</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>E.A.</td>
<td>z</td>
<td>E.A.</td>
<td>z</td>
</tr>
<tr>
<td><strong>Personal Skills</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Interaction with adults</td>
<td>-0.52</td>
<td>-1.08</td>
<td>-0.36</td>
<td></td>
</tr>
<tr>
<td>Expression of feelings/affect</td>
<td>-1.75</td>
<td>-2.05</td>
<td>-2.91</td>
<td></td>
</tr>
<tr>
<td>Self-concept</td>
<td>-1.40</td>
<td>-1.28</td>
<td>-1.65</td>
<td></td>
</tr>
<tr>
<td>Interaction with peers</td>
<td>-1.18</td>
<td>-1.75</td>
<td>-1.84</td>
<td></td>
</tr>
<tr>
<td>Coping</td>
<td>-0.92</td>
<td>-0.64</td>
<td>0.01</td>
<td></td>
</tr>
<tr>
<td>Social Role</td>
<td>-1.76</td>
<td>-0.71</td>
<td>0.71</td>
<td></td>
</tr>
<tr>
<td><strong>Total Personal Skills</strong></td>
<td>44</td>
<td>-1.23</td>
<td>52</td>
<td>-1.40</td>
</tr>
<tr>
<td><strong>Adaptive Skills</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Attention</td>
<td>-0.50</td>
<td>-0.95</td>
<td>0.40</td>
<td></td>
</tr>
<tr>
<td>Dressing</td>
<td>-7.13</td>
<td>-1.86</td>
<td>-0.41</td>
<td></td>
</tr>
<tr>
<td>Personal responsibility</td>
<td>-0.92</td>
<td>-0.84</td>
<td>-0.36</td>
<td></td>
</tr>
<tr>
<td>Toileting</td>
<td>-2.05</td>
<td>-2.05</td>
<td>-0.61</td>
<td></td>
</tr>
<tr>
<td><strong>Total Adaptive Skills</strong></td>
<td>45</td>
<td>-1.28</td>
<td>54</td>
<td>-1.04</td>
</tr>
<tr>
<td><strong>Motor Control</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Body coordination</td>
<td>-1.64</td>
<td>-3.89</td>
<td>-3.89</td>
<td></td>
</tr>
<tr>
<td>Locomotion</td>
<td>-1.97</td>
<td>-3.78</td>
<td>-3.78</td>
<td></td>
</tr>
<tr>
<td>Coarse Motor Control Total</td>
<td>16</td>
<td>-3.28</td>
<td>39-42</td>
<td>-4.58</td>
</tr>
<tr>
<td>Fine muscle control</td>
<td>-1.64</td>
<td>-1.04</td>
<td>-0.27</td>
<td></td>
</tr>
<tr>
<td>Perceptual motor control</td>
<td>-0.44</td>
<td>-1.75</td>
<td>0.17</td>
<td></td>
</tr>
<tr>
<td>Fine Motor Control Total</td>
<td>46</td>
<td>-0.88</td>
<td>57-58</td>
<td>-1.64</td>
</tr>
<tr>
<td><strong>Total Motor Control</strong></td>
<td>31</td>
<td>-2.61</td>
<td>49</td>
<td>-3.24</td>
</tr>
<tr>
<td><strong>Communication Skills</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Receptive</td>
<td>54</td>
<td>0.00</td>
<td>65-66</td>
<td>-0.18</td>
</tr>
<tr>
<td>Expressive</td>
<td>50</td>
<td>-1.88</td>
<td>61</td>
<td>-0.84</td>
</tr>
<tr>
<td><strong>Total Communication Skills</strong></td>
<td>51</td>
<td>-0.25</td>
<td>62</td>
<td>-0.61</td>
</tr>
<tr>
<td><strong>Cognitive</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Perceptual discrimination</td>
<td>-0.44</td>
<td>-0.74</td>
<td>0.78</td>
<td></td>
</tr>
<tr>
<td>Memory</td>
<td>-0.39</td>
<td>-1.00</td>
<td>-0.76</td>
<td></td>
</tr>
<tr>
<td>Reasoning and academic skills</td>
<td>-0.61</td>
<td>-1.88</td>
<td>-0.27</td>
<td></td>
</tr>
<tr>
<td>Conceptual development</td>
<td>0.05</td>
<td>-0.28</td>
<td>1.00</td>
<td></td>
</tr>
<tr>
<td><strong>Total Cognitive</strong></td>
<td>53</td>
<td>-0.23</td>
<td>59</td>
<td>-0.92</td>
</tr>
<tr>
<td><strong>Inventory Total</strong></td>
<td>47</td>
<td>-1.04</td>
<td>57</td>
<td>-2.05</td>
</tr>
</tbody>
</table>
discrimination, reasoning and academic skills and conceptual development; the memory score was slightly lower, but still within the normal range for this age group.

In 1996, at the age of 56 months, the boy began to attend an ordinary infants school. This had a very positive influence in most areas of his development, especially as regards socialisation: the total indifference he showed towards peers during the first few months gradually disappeared, and he currently has a group of friends with whom he is at ease and also relates better than before with other class members. At the same time, his attitude to school work also improved from initial apathy to the expression of interest for all school activities. The only developmental areas in which schooling has had a negative influence are self-esteem and self-confidence, which at the start of schooling were fairly acceptable but then declined sharply.

To sum up, this boy’s development has exceeded all initial progress. He is well-integrated, pacific and reasonable, and responds satisfactorily to all tasks set. He continues to attend the Early Intervention Unit and to receive speech therapy with a view to both monitoring his progress and improving those aspects of development in which he still lags behind his peers.

Appendix

A SAMPLE PERSONALISED PROTOCOL

Successive protocols were drawn up roughly every 3 months, although the exact duration of each depended on the subject’s performance. Objectives and activities were selected from the lists provided in the Portage Guide to Early Education (Bluma et al., 1972), and were adapted appropriately if necessary.

The objectives pursued changed as the subject acquired new skills; no objectives were set that depended on skills not already acquired.

Protocol for D.M.L. at 18/12/96

Chronological age: 2 years 11 months

Course Motor Control

Objective 1. Jumping with feet together
Stand in front of D, holding him by both hands. Make him bend his knees as you do. Lift his hands up as you jump with your feet together.
Make him climb to the first step of a flight of stairs or toy step-ladder and turn round to face you. Hold both his hands and help him to jump.
Progressively reduce the help you give him.

Objective 2. Walking down stairs with help.
Make him practice walking down stairs. Start from the bottom step, and gradually increase the number of steps.
Face him on the stairs, coming down backwards as he comes down forwards.
Encourage him to place one foot on the next step down.
Lift his foot and place it on the next step down.
Reward him every time he tries to come down a step.

Objective 3. Walking backwards
Work in a large clear area to avoid D bumping into anything. Show him how to walk backwards.
Help him to walk backwards by standing behind him and pulling him backwards.
Stand in front of him, hold his hands, and move one of your feet slowly forward so that it prods the toe of his foot to prompt him to move it backwards. Then do the same with the other foot, and so on.
Reward him and say ‘back’ every time he steps backwards.

Fine Motor Control

Objective 1. Building a tower of 5 or 6 blocks.
Build a six-block tower in front of D.
Give him another six blocks and ask him to build a tower.
Praise him every time he adds a block to the tower. If he stops, give him another block; if necessary, guide his hand. Praise him if he does well. Let him knock the tower down as a reward. Make him practice with other materials, such as sponges, boxes, etc.

Objective 2. Turning the pages of a book one by one.
Give D some books to look at and handle. Make him turn the pages as you read him a story. Praise him when he does so. Use a picture book with cardboard pages. Show him a photo of himself. Place it between the pages of a book. Encourage him to turn the pages to find the photo. Make an album of pictures of his favourite things. Stick tabs on the pages so they are easy to turn.

Socialisation
Objective 1. Fetching an object or a person to or from another room when told to do so.
Make D call his father or his brother at meal times. Send him to do simple errands (Take daddy the newspaper, Fetch a nappy etc.); thank him for his help. At first, go with him to help him do the errand. Later, let him go alone, following verbal instructions. Praise him when he does it properly. At first, give very simple instructions. Later, make the errands gradually more complex.

Objective 2. Paying attention to music or stories for 5-10 minutes.
Read D a simple, interesting story, explaining the pictures to him. Ask him to tell you what he sees in them. Ask him simple questions about the story to find out whether he was paying attention. Read to him every day, preferably at the same time each day. Make this an enjoyable part of the day for him. Let him choose the story. Play a recording of a tune or song forming part of some simple music-and-movement game. Play the game with D. Later, watch to see whether he plays the game by himself when he hears the music.

Objective 3. Helping with chores.
Encourage D to help at home. First, make him responsible for putting his toys away. Praise him when he does. Let him take his plate to the sink. Let him help you change the bedclothes, put the groceries in their place, etc. Thank him and praise him for helping. Let him hold the dustpan when someone is sweeping.

Language
Objective 1. Combining word and gesture to express wishes.
Choose an activity that D likes, such as playing ball. If he says ‘ball’, ask ‘Who wants the ball?’ Point to him and say ‘ball’. Wait for him to point to himself and say ‘ball’ before throwing it to him. If he doesn’t, take his finger and make him point to himself, and say ‘ball’ to him; then throw the ball. Repeat this sequence of waiting and instruction. When he knows what is expected of him, wait until he points to himself and says ‘ball’ before throwing it to him. Use the same procedure when he plays at blowing bubbles, for example. Use toys such as a toy farm or a doll’s house. Set up the farm or the doll’s house, but keep back all except one or two animals, dolls, pieces of toy furniture, etc. Let him see that you have the rest. If he holds his hand out, ask him what he wants. Pronounce the name of an animal or doll clearly if he is unable to do so. Encourage him to say his own name, or ‘me’, and to point to what he wants. When he understands what is expected of him in these games, gradually start to make him do the same to ask for his everyday needs to be satisfied. Don’t let him go out, get a biscuit or drink, or play with his favourite toy until he lets you know what he wants, combining word and gesture. Start demanding this in three or four specific situations, and then include more.

Objective 2. Naming five members of the family (including pets if appropriate).
Use the name of each member of the family frequently. Encourage D to distinguish among different members of the family, saying, for example, ‘Give Mama the ball’.
Get the members of the family to ask him their names. A good time to practice is at meal times.

Self-Help
Objective 1. Sitting on the potty for 5 minutes. When you are changing his nappy, talk to him about using the potty, doing pee, or whatever; use the expression you like, but always use the same. Let him see members of the family using the lavatory. Try to make sure that everyone uses the same expression, the one you are teaching him.
Sit him on the potty. Stay with him and praise him for staying seated. Acustom him to it by telling him a story while he is seated.
Sit him on the potty twice a day at times when he is likely to urinate. Stay with him and praise him if he does urinate.
Make this a pleasant event for him so as to encourage him to stay seated. This first stage in toilet training is important for the formation of good habits. Don't demand that he remain on the potty for more than 5 minutes at a time.

Cognition
Objective 1. Distinguishing big from little.
Collect groups of big and little examples of the same kind of object (plates, blocks, pencils, biscuits, etc.)
Place a big pencil, a little pencil and a sheet of paper in front of D. Ask him to draw a line on the paper with the big pencil. Praise him if he does. Repeat analogous activities with other kinds of objects.
Ask D to find big and little objects
Play motor development games such as taking big steps and little steps forward, sitting in a big chair and a little chair, etc.

Objective 2. Placing objects in, on top of or under others when asked.
Use a small block and an upside-down cup. Tell D you are going to put the block under the cup. Make him imitate you. Play analogous games to teach him 'on top of' and 'in'.
Get a big, sturdy cardboard box and ask him to get in it, on top of it, and under it.
Take one of his favourite toys and place it in, on top of and under a box. Then encourage D to place the toy the way you ask him to. Praise him when he gets it right. Give him all the help he needs, reducing the amount of help as his competence becomes consolidated.

Objective 3. Naming action shown in pictures.
When you read D stories, point to the pictures and tell him what they show.
Play guessing games with him. Play-act actions such as crying, running, driving, laughing, etc., and make him tell you what you are doing.

Summary
A child with Potter’s sequence due to unilateral renal agenesis was given stimulation treatment at home and in our Early Intervention Unit since age 23 months. By age 65 months his development had recovered notably in all areas, allowing successful access to normal schooling.

References
pulmonary hypoplasia (Potter’s syndrome). Investigation & Radiology, 17, 6-10.