Intern. J. Neuroscience, 116:361–369, 2006

ISSN: 0020-7454 / 1543-5245 online
DOI: 10.1080/00207450500455330

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Taylor & Francis Group

A NEW SYNDROME WITH QUADRUPEDAL GAIT, PRIMITIVE SPEECH, AND SEVERE MENTAL RETARDATION AS A LIVE MODEL FOR HUMAN EVOLUTION

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The author has discovered a new syndrome with quadrupedal gait, flexed head and body, primitive speech, severe mental retardation, and mild cerebellar signs with a disturbed conscious experience. This syndrome was exhibited by 5 of 19 children from a consanguineous family. The pedigree demonstrated a typical autosomal-recessive inheritance. The genetic nature of this syndrome suggests a backward stage in human evolution, which is most probably caused by a genetic mutation, rendering, in turn, the transition from quadrupedality to bipedality. This would then be consistent with theories of punctuated evolution. On the other hand, the extensor motor system causing a resistance of the body against the gravity may actually be subjected to evolutionary forces. This new syndrome may be used as a live model for human evolution.

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Keywords erect posture, evolution, human quadrupedal gait

Received 1 August 2005.

The author thanks Dr. Osman Demirhan for introducing the family, Dr. Meliha Tan for the neurological examinations, and Derya Deniz Elalmis for performing the peg-moving task.

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INTRODUCTION

The present work describes a new syndrome, and its relation to human evolution, especially with regard to transition from the habitual quadrupedality to habitual bipedality.

As generally known, human beings are unique in articulated speech, habitual bipedality, and high intelligence as well as the conscious experience. The transition from quadrupedality to bipedality, that is, the ability to walk habitually on two legs, is the most important stage in human evolution. This is taken as the most important, first step, predating the other uniquely human traits.

The emergence of an upright posture suggests a psychological resistance —resistive mind—of our ancestors against the very strong gravitational forces of earth on which they live. The resistive mind apparently co-evolved with increasing brain size. A large brain in our ancestors seems to be coupled with resistive behavioral traits, which were probably the triggering factors for the emergence of homo erectus, that is, habitual upright posture with bipedal gait. Thus, the author maintains that there was first the free will as a main trait of the large brain, equipped with an intelligent struggle for human survival. The resistive human mind against the earth‘s gravitational forces is still in action, despite having the habitual erect posture since millions of years. As a result, not only are humans habitually standing up on two feet, they are even completely freed from the gravitational forces, and walking within the depths of the endless space.

In light of these considerations, this article describes a new syndrome with quadrupedal gait, primitive speech, severe mental retardation, and disordered conscious experience. This syndrome, apart from being a rare neurological disease, may elucidate the human evolution, with gradual or punctuated emergence of human beings.

METHOD

The participants originated from a consanguineous family in a small village in Southern Turkey. The father (65 years old) and mother (56 years old) were relatives, but not very close. They had 19 children, which are now between 14 and 36 years of age. Twelve of their children exhibited bipedal gait, two died early. Five children exhibited quadrupedal gait, that is, they walked on two hands using their palms, and two feet with straight legs. The family tree demonstrated a typical autosomal-recessive inheritance of a disorder with a transmission by affected males and females to the offsprings

of both sexes. According to the mother‘s report, the affected children were born after normal gestation and delivery. They have never acquired an erect posture during childhood, but began to walk on four extremities at about three or four years of age.

Upon physical examination, mild thoracal scoliosis was present. Upon neurological examination, they were awake, the cranial nerves were intact, muscle tonus and tendon reflexes were mildly decreased; there were no extrapyramidal signs and symptoms. They had bilateral dysmetria and dysdiadochokinesis. Muscle weakness, sensory loss, and deformities were not observed. Cranial magnetic resonance imaging revealed mild cerebral and cerebellar atrophy (axial T1 = weighted images) in quadrupedal participants. Otherwise, no abnormalities were seen in MRI and the whole-body CT examinations.

The intelligence and consciousness level of the participants were assessed using the “Mini Mental State Examination Test,” standardized for the uneducated Turkish people. The total score is 30 points. Scores in the range of 0–23 indicate a disturbance of cognition. This test attempts to measure the patients capabilities in five fields: orientation (date and location), registration (immediate recall of three words), attention and calculation (count backwards), recall (recall three items), and language and drawing (name a few items, repeat a sentence, paper folding, draw a watch).

RESULTS

The date of examination was May 1, 2005, in the morning; the country they live in is Turkey.

Sitting Posture

The sitting postures of the quadrupedal children are shown in Figure 1. As seen in this photograph, they could not hold their heads upright due to scoliosis. Their heads were flexed forward with their skulls, which also was flexed forward on their spines. They could not raise their heads to look forward. This forward head posture with flexed skull is quite similar to the head posture during standing in our closest relatives, like chimpanzees.

Standing Posture

The quadrupedal individuals occasionally stood up for a few seconds. However, this was not a fully erect position. A representative example is shown in Figure 2. As seen in this photograph, if they wanted to stand up from time to

Figure 1. Sitting positions in quadruped patients. Notice the forward flexed heads. (See Color Plate XI at end of issue.)

time for unknown reasons, their bodies with their legs were in a flexed position while standing up. With these features their upright posture was rather similar to our closest relatives, the chimpanzees.

Walking

Figure 3 illustrates the quadrupedal walking patterns of the patients. Their quadrupedal gait was similar to diagonal walking seen in many animals, such as dogs, horses, chimpanzees. As seen in the photographs, in Figure 3, the feet at diagonal ends of the body strike the ground together in diagonal gait. The balance and support are maintained, for instance, by the left foot-right hand while the left hand—right leg are suspended, and then the opposite diagonal starts for further walking action. Similar to most primates, they habitually used diagonal sequence of footfall pattern, in which the footfall of a foot was followed by that of a contralateral hand. The patients exerted nearly equal weights to the palms and feet during the quadrupedal walking, as measured by four balances for two palms and two feet. They could walk fairly fast with strong legs, without any imbalances and ataxic movements.

Figure 2. Representative standing position in one of the quadruped patients. (See Color Plate XII at end of issue.)

Speech

The patients had articulated speech, although most of the words were not understandable. They had their own language to communicate and chat with each other. Their mother and father could rather easily communicate with them, but not the other people. These quadrupeds used less than a hundred words to express their wishes.

Intelligence

Using the Mini Mental Examination Test, the questions and the answers of the patients are presented in Table 1. The patients always had zero points in

Figure 3. Samples for the quadrupedal gaits. Notice the diagonal walking patterns. (See Color Plate XIII at end of issue.)

Table 1. Questions and answers of the quadruped individuals used in MMSE test

Questions	Patient's answers
What is the year?	80, 90, animals, july, house
What is the season?	Summer, me, winter, animals, mother
What is today?	No reply, celebration, 19th, no reply, father
Morning, noon, afternoon?	Afternoon, morning, evening, tomorrow, afternoon
Repeat: apple, table, money	They could not repeat
Which country are we living in?	Germany, no reply, America, no reply, no reply
Which village are we living in?	Nobody, no reply, Demirkonak, me, no reply
Count backwards from10 to 1!	Nobody could count
Count forwards from 1 to 10	Nobody could count
Recall the names: tish, flag, clothe	Nobody could recall
What is this (red shoe)?	Tomato, shoe, no reply, melone
What is this (watch)?	Watch, no reply, glass, no reply, no reply
What is this (book)?	No reply, book, no reply, no reply, no reply
Repeat the phrase (no ifs, ands, buts)	Nobody could repeat
Take the paper, fold it in half once	They only played with the paper

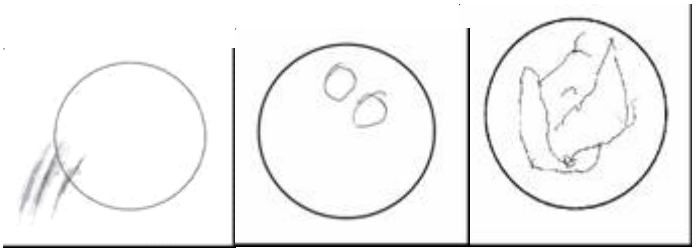

Figure 4. The results of draw-a-watch test (within the circle) in three patients.

this test, except one who had 2 points. The patients were also unable to draw a watch. Figure 4 illustrates the results of the draw-a-watch-test in three patients.

DISCUSSION

As described earlier, the author has discovered a new syndrome with quadrupedal palm-gait, severe mental retardation, primitive speech, and disturbed conscious experience. This is essentially an inborn gait disorder because the patients have never acquired an erect posture and bipedal gait after childhood. Apart from being a very rare disorder, this syndrome interestingly exhibits prehuman features. This new syndrome can be considered a live model for human evolution. In this respect, the quadrupedal walking is a most interesting trait.

It is generally accepted that human beings are unique in habitual erect posture, bipedal gait, articulated speech, and conscious experience with high intelligence and a large brain. There is much debate about the transition from the habitual quadrupedal gait in our closest cousins, the African apes, to the habitual bipedal gait in humans. It is, however, not known whether the transition from our common ancestors to human beings occurred slowly by gradual evolution or by sudden mutation or mutations in special genes (punctuated evolution). Now, the new syndrome, which may be called thereafter as "Uner Tan Syndrome" considering the discoverer's name, suggests a punctuated human evolution. That is, the transition from the habitual quadrupedality in our common ancestors to the habitual bipedality in human beings did not occur through a gradual evolution; the homo erectus with bipedal walking pattern and large brain may have emerged by sudden mutation(s) in special genes. The new syndrome shows a backward step in the walking pattern, that is, from bipedality in humans to quadrupedality in our closest cousins. In other words, the same specific mutation causing a quadrupedal walk in quadruped patients may also be involved in transition from quadrupedality to bipedality.

So, the erect posture of human beings may be the result of a punctuated evolution by invoking special gene(s). The origins of standing up from horizontal to vertical position may actually be the extensor motor system. That is, the evolution of the bipedal gait may not be the walking style itself, rather the evolution of the extensor motor system may indeed play the main role in the origins of human uniqueness. The extensor motor system mainly involves the skeletal muscles that keep the body upright against gravity. To be able to stand up despite the very strong gravitational forces, one must first have a brain equipped with a resistive mind. This is the human mind. The flexor motor system was dominant in the quadruped patients: their heads and bodies were flexed during sitting and occasional standings, similar to our ancestors, the African apes, which do not have a resistive mind, and extensor dominance. Thus, according to this scenario, there was first the relatively large human brain exhibiting a resistive human mind, which forced humans to resist the gravitational forces, and stand up.

The newly discovered syndrome with a possible backward evolution due to a genetic mutation suggests a co-evolution of large brain with human mind, articulated human speech, high intelligence, conscious experience, and habitual erect position with bipedal gait. Taking this syndrome as a live model of human evolution, it can be concluded that these traits making them unique among other animals may be co-evolved as a result of a punctuated genetic event including sociocultural factors, such as the influence of the most intelligent individuals. The disordered cognitive abilities with a gait disorder in the patients due to a genetic anomaly support the notion that all of the human characteristics may have evolved together. Accordingly, the patients had severe mental retardation with a very primitive consciousness and speech abilities in addition to quadrupedal walking pattern.

The patients had a habitual walking pattern, which may be called a palm walking, because they walked on their palms; some of them raised the fingers at the same time. Contrarily, our close relatives usually exhibit a knuckle-walking. The palm-walking in the quadruped individuals does not support the notion that our ancestors were also knuckle-walkers, and humans lost it during evolution. If so, it seems to be possible that the knuckle-walking was changed to palm-walking just prior to upright bipedal-walking. Thus, the palm-walking may throw some light on the unresolved problem about the walking pattern in our ancestors just prior to modern human beings.

Interestingly, there was a severe mental retardation coupled with a very primitive conscious experience in addition to a postural gait disorder. This supports the author's idea that there was first a resistive human mind coupled with a well-developed extensor motor system. According to the "psychomo-

tor theory" (see Tan, 2005), the extensor motor system is closely coupled with human mind, and this motor nucleus is the mind itself at the same time, expressed by human language, which also included in the motor system. According to the results of the Mini Mental State Examination, the patients were unaware of time, place, and date. They simply lived without any interest for the world in which they lived. Thus, it is conceivable that the extensor dominance, especially active in waking states, may be coupled with the human mind with conscious experience. The expressive part of the human motor system was also impaired in these patients: they usually talked to each other, but nobody could understand them except their father and mother. They had a very limited vocabulary. They could not even name several everyday items (see Table 1). They did even not know where they are living, in which country, in which village, which year, and so on. So, their consciousness also stayed in a primitive level. These findings also suggest that coupled with a quadrupedal gait and flexor tonus instead of an extensor tonus, the mental abilities also stayed in a very primitive level. Such a syndrome must be very rare, or the first and the last one, but this may be a unique live model to have some ideas about how our ancestor(s) just lived prior to modern humans.

CONCLUSIONS

The present work described a new syndrome exhibiting a habitual palm-gait, primitive speech, severe mental retardation, and severely impaired conscious experience. This genetic disorder suggests a backward stage in human evolution, following a sudden mutation(s) (punctuated evolution), not resulting from a slowly occurring gradual evolution. Accordingly, this new syndrome suggests, as a live model for human evolution, that first of all a human-specific resistive mind coupled with a relatively large human brain emerged during the human evolution. These human beings resisted the earth's gravitational forces as a result of the evolution of the extensor motor system, which was the actual driving force for humankind including the upright posture against gravity. This psychomotor drive is still in action, causing humans to be completely free from the earth‘s gravitational forces, even reaching the deepness of endless space.

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