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EVIDENCE FOR "UNER TAN SYNDROME" AS A HUMAN MODEL FOR REVERSE EVOLUTION

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"Uner Tan Syndrome" was further studied in a second family. There was no cerebellar atrophy, except a mild vermial atrophy in MRI scans of the affected individuals. This is not, however, the pathogenesis of the "Uner Tan Syndrome", since in the first and second families there were bipedal men exhibiting very similar MRI scans. The second family may also be considered a live model for reverse evolution in human beings. The present work provided evidence for a reverse evolution: (i) quadrupedality; (ii) primitive mental abilities including language; (iii) curved fingers during wrist-walking of the quadrupedal woman; (iv) arm to leg ratios being close to those of the human-like apes. The quadrupedal individuals were raised in separate places, so that they could not imitate each other, excluding the socio-cultural factors contributing to the habitual quadrupedal gait. The results are consistent with the single gene theory, suggesting a single gene controlling multiple behavioral traits, and the psychomotor theory, and a co-evolution of the human mind, an emergent property of the motor system expressed by human language.

Key words: Uner Tan Syndrome, quadrupedal gait, reverse evolution, cerebellum, intelligence, speech, gait

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INTRODUCTION

"Uner Tan Syndrome" consists of three main symptoms: quadrupedal gait, mental retardation, and primitive speech (see Tan, 2005b; Tan, 2006a, b). The mini mental state examination test standardized for uneducated Turkish individuals indicated a low-level conscious experience in addition to a severe mental retardation. The first examination of the family was May 1st, 2005 (see Tan, 2006a, received 1st August 2005).

The mental abilities of the affected individuals were not assessed by others (Humphrey et al., 2005; Turkmen et al., 2005), although they have claimed that these individuals were mentally retarded. However, using the mini mental state examination test standardized for the uneducated Turkish adults, Tan (2005b, 2006a, b) showed that they had severe mental retardation with disturbed conscious experience. The sitting and standing postures were also analyzed only by Tan (2005b, 2006a, b). Interestingly, they exhibited flexor posture during sitting and standing, similar to our closest relatives, such as bonobos and chimpanzees. The language abilities of the affected individuals within the first and the second families were also limited to a primitive level.

Humphrey et al. (2005) stated that "it is a syndrome that may never be seen again". Contrary to this scientifically not substantiated prediction, I have discovered sporadic cases in two other Turkish cities far from each other, and further individuals exhibiting the "Uner Tan Syndrome" in another Turkish family, also with intra-familiar marriage as in the first family (see Tan, 2006b). There were actually four affected individuals within the second family: one quadruped man, one quadruped woman, one biped man with ataxic gait, and one totally handicapped man with no walking ability at all. The biped ataxic man and the latter were also quadrupeds during their childhoods, while the first one developed to bipedality during adulthood, and the latter one progressively worsened to become a totally handicapped person. These individuals were not siblings; they were close relatives, raised in separate places so they could not imitate each other to become a quadruped, contrary to Humphrey et al. (2005). The present work deals with a detailed analysis of the second family especially with regard to the walking patterns, MRI scans, and bodily measurements in relation to the theory of reverse evolution (see Porter & Crandall, 2003).

METHODS

The participants of the present study belong to the second family discovered by Tan (2006b). There was one quadruped man (37 years of age) and one quadruped woman (27 yeas of age) as seen in the pedigree of this family shown

in the previous article (see Tan, 2006b). There were intra-familiar marriages within the last family, as in the first family.

Upon neurological examination they were awake, the cranial nerves were intact, muscle and tendon reflexes were normal; there were no extrapyramidal signs and symptoms. They had bilateral dysmetria and dysdiadochokinesis. No muscle weakness or sensory loss were observed. They had very low-frequency intentional tremor and mild horizontal nystagmus.

T1-weighted Cerebral MRI scans were made, to see abnormalities in the cerebral and cerebellar structures, using the coronal and sagittal sections of the 5 mm-thick brain scans.

The weight, height, and the lengths of their arms and legs were measured, and the ratio of the arm- to leg-length was calculated.

According to the mother's report, the affected children were born after normal gestation and delivery. Their crawling was normal, as seen in the majority of babies, however, they could not stand up and walk on two legs after the crawling period.

The intelligence level and conscious experience were assessed using the "Mini-Mental State Examination Test", standardized for the uneducated Turkish adults. The total score was 30 points in this test. The test attempts to measure the 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 (name a few items, repeat a sentence). I have taken a written consensus of the family for every investigation on them. In addition, I have taken an approval form the Ethics Committee of the Medical School, Cukurova University.

RESULTS

Bodily Measurements

The height and weight of the quadruped man was 147 cm and 86 Kg, respectively. The weights exerted during quadrupedal position were found to be 5 Kg for the right hand, 10 Kg for the left hand, 26 Kg for the right foot, and 39 Kg for the left foot. The lengths of the arms and legs were found to be 67 and 70 cm, respectively. The arm to leg ratio was found to be 0.90. This ratio was 0.83 and 0.77 for the bipedal man with ataxia and the quadrupedal woman, respectively. It was 0.68 for the author (UT) and 0.60 in a journalist woman.

Fist-Walking

The quadruped man used his palms to touch the ground, while the quadruped woman used her fists to touch the ground during walking. The fingers of the

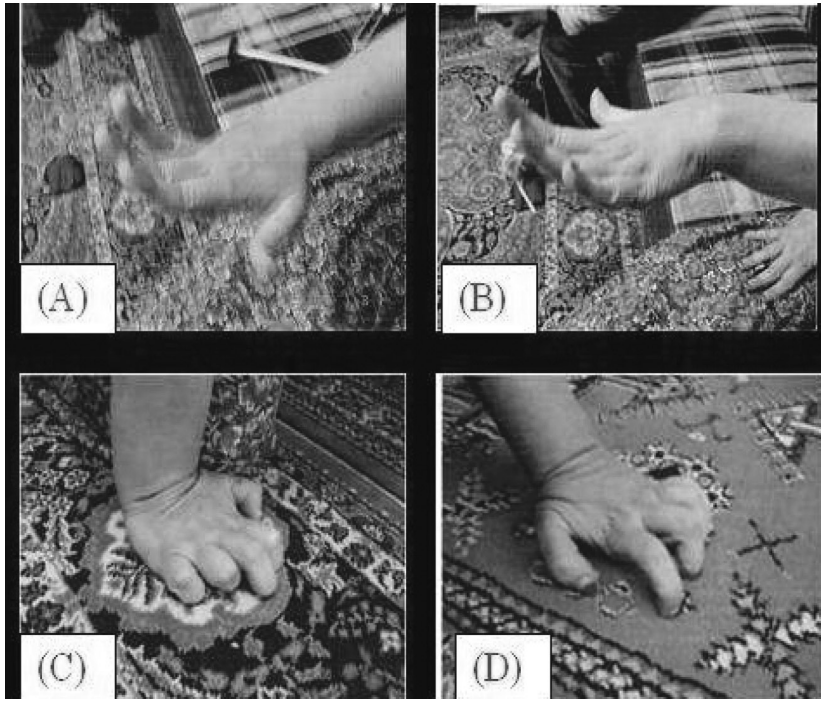

Figure 1. Wrist walking with flexed (curved) fingers of the quadruped woman. Above, the hyperextended fingers during stretching of the right (A) and left (B) arms; Below, the flexed fingers of the right (C) and left (D) hands during quadrupedal walking. (See Color Plate V)

quadruped woman were flexed (curved) during quadrupedal walking. Figure 1 illustrates the hands of the quadrupedal woman during walking (C: right hand, D: left hand). As she stretched her hands during sitting, the fingers were hyperextended (see Figure 1, A: right hand; B: left hand).

Brain Scans

The quadruped man and woman as well as the biped man with ataxic gait exhibited similar MRI scans. That is, their cerebellum seemed to be normal, while there was a mild vermial atrophy in all of them including the quadruped woman (see Figure 2).

Neurological Examination

As seen in the first family, there were mild cerebellar signs and symptoms in all of the affected individuals: dysmetria, dysdiadochokinesis, mild horizontal

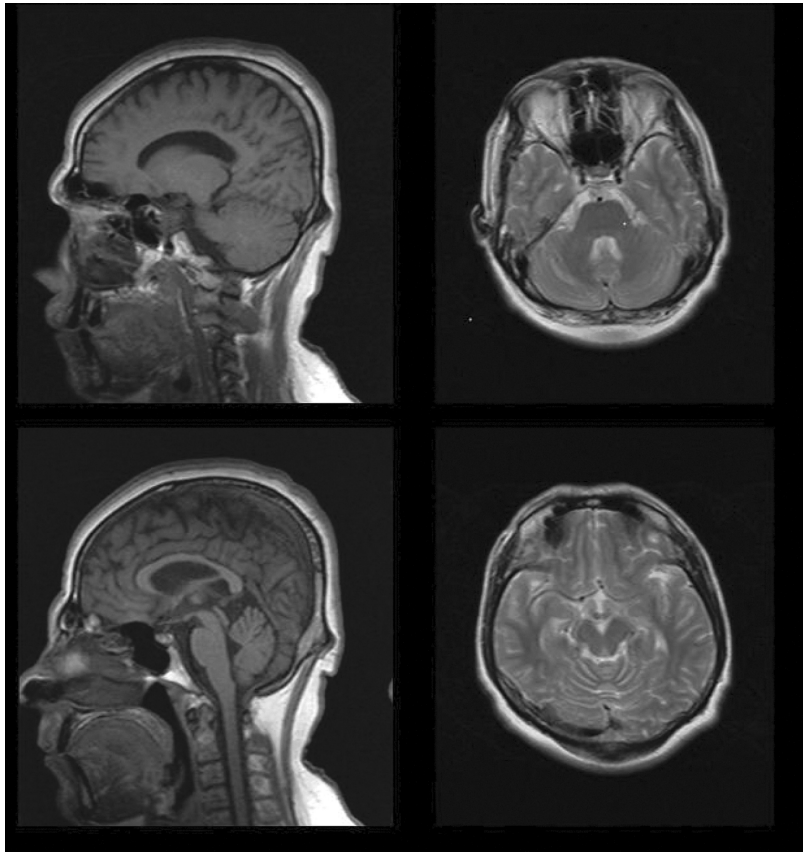

Figure 2. MRI scans for the quadruped man. Left: sagittal sections, above normal cerebellum, below mild vermial atrophy; Right: coronal section, above normal cerebellum, below mild atrophy in cerebellar vermis.

nystagmus, very low-frequency intentional tremor, and an ataxic gait in bipedal man. Otherwise, they were awake, the cranial nerves were intact, muscle tonus and tendon reflexes were normal; there were no extrapyramidal signs and symptoms. No muscle weakness and sensory loss were observed.

Hand Skill

The affected individuals (two quadrupeds, one biped with ataxic gait, and one totally handicapped) were very slow and inaccurate in hand skill measured by the peg moving task, generally used to assess the hand skill. They could not bind

their shoes themselves. The quadrupedal woman could not bind her head cover herself. They used their right hands to eat, while they had great difficulty in reaching their mouths, because of the very low-frequency and large-amplitude tremor in their hands.

Speech

Their speech was very slow and not easily understandable because of the difficulties in articulation. They used a limited vocabulary and very short sentences consisting of only a few words, to communicate with others. They did not use the words "and", "with". They always used very simple sentences consisting of a few words. They could not name many objects.

Intelligence

The "mini mental state examination" test was used to assess their cognitive abilities. This test measures the capabilities in five fields: orientation (date and location), registration (immediate recall of three words), attention and calculation (count backwards and forwards), language (recall three items, repeat a sentence, name a few objects shown from a picture book). None of the tested individuals achieved a score greater than zero. They could even not say the name of their father or mother. They were unaware of day, time, place, season; they could not count from 1 to 10 either.

DISCUSSION

This study provided evidence for the "Uner Tan Syndrome", and a reverse–backwards-evolution. The three main symptoms of the "Uner Tan Syndrome", i.e., the quadrupedal gait, severe mental retardation, and primitive speech, could be clearly substantiated in the individuals of the second family. Similar to the first family, this family also suggested that this syndrome has genetic origins exhibiting an autosomal recessive inheritance (see Tan, 2006b).

"Uner Tan Syndrome" cannot merely be explained by a cerebellar atrophy, since the cerebellum was normal as detected in MRI scans of the affecetd individuals: qaudrupedal man, quadrupedal woman, and bipedal man with ataxic gait, being also quadrupedal during childhood. There was a mild vermial atrophy in all of the affected individuals, however, this cannot explain the origins of the "Uner Tan Syndrome." There are too many cases in the scientific

literature pointing out a cerebellar and vermial hypoplasia, but none of them described a quadrupedality. Growing up in separate environments and sporadic cases seen in different regions of Turkey far from each other excludes any socio-cultural influences on the development of the quadrupedal gait, contrary to Humphrey et al. (2005).

Interestingly enough, the quadrupedal woman used flexed (curved) fingers during quadrupedal walking. This is of course not a knuckle walking, such as in chimpanzees, and fist-walking as in orangutans, but it may be a trace of the knuckle walking or fist-walking as a result of a reverse evolution due to a genetic defect. The same defective gene, causing a reverse evolution from bipedality to qudrupedality, may be the gene undergoing a favorite mutation during the transmission from quadrupedality to bipedality in the evolutionary history of human beings.

Interestingly enough, the ratio of arm length to leg length was found to be 90% for the quadrupedal man, 83% for the bipedal man with ataxia, and 78% for the quadrupedal woman. Human-like apes have a ratio about 90% or slightly more, Lucy's ratio was estimated to be 85%, and human have a ratio of around 70% (see e.g., Korey, 1990). The ratio of the arm- to leg-length found in the affected individuals, especially that for the quadrupedal man, being 90%, support the theory of a backward evolution. So, not only a single trait, the quadrupedal gait, but many other features, such as the primitive language, primitive intelligence, primitive conscious experience, fist-walking, and the arm to leg ratios, all of them support the theory of devolution (reverse evolution) or a backwards evolution in the individuals exhibiting "Uner Tan Syndrome". This is the first human evidence for the reversed evolution. Otherwise, there are reports for the evidence for the reverse evolutions in plants and animals (see Porter & Crandall, 2003).

There is a possibility to find the single gene playing a role in the transition from quadrupedality to bipedality including all of the co-evolved human traits, such as the speech and intelligence including the human mind, if we can find the responsible gene for the reverse evolution. In accord, some scientists provided experimental evidence that a single gene can influence the complex social and sexual behaviors (e.g., Manoli et al., 2005; Wahlsten, 1999). Accordingly, a single gene might be responsible for the co-evolution of the unique human properties such as bipedal gait, high intelligence, and language including human mind, which may be an emergent property of the motor system expressed by human language, according to the "psychomotor theory" (Tan, 2005a).

The "Uner Tan Syndrome" can be considered a live model for the punctuated evolution. In this context, some hotspots were found in human

genome for acquiring duplicated DNA sequences—but only at specific time-points during evolution. It seems that long periods of genomic stasis are punctuated by relatively short episodes of duplicate activity; new DNA repeats arise in the genomes of species in irreversible and punctuated events (Minghetti & Dugaiczyk, 1993). The debate on long-term rates of evolution is accumulated around two models: the punctuated equilibrium hypothesis and the gradual thesis, the latter being a gradual rate of evolution, and the former one being long periods of very little evolution interrupted by periods of relatively rapid evolutionary changes. During evolution, beneficial mutations can sweep successfully through the population, giving rise to punctuated evolution (Elena, Cooper, & Lenski, 1996). Mezhzherin (1997) has analyzed the genetic differentiation of taxa from three Holarctic and three Afrotropical phyla of small mammals, and found that (i) the distributions of fixed gene differences were relatively independent; (ii) the speciation process was discontinuous. These were consistent with punctuated equilibria, i.e, a discontinuity in speciation process.

CONCLUSIONS

I have further presented the second family as evidence for the "Uner Tan Syndrome". There was no cerebellar atrophy, except a mild vermial atrophy in MRI scans. This cannot, however, account for the origins of the "Uner Tan Syndrome", since, in the first and second families, there were bipedal men exhibiting very similar MRI scans, and there are no quadrupedality in cerebellar ataxias frequently reported in the scientific literature. As stated in the previous article (Tan, 2006b), the autosomal recessive transmission may indicate a genetic mutation responsible for a reverse evolution first time in the human history. Accordingly, the present results provided evidence for a reverse evolution in the individuals exhibiting the "Uner Tan Syndrome": (i) quadrupedality; (ii) primitive mental abilities including language; (iii) curved fingers during wrist-walking, (iv) the ratios of the arm- to leg-lengths, close to human-like apes. The quadrupedal individuals were grown up in separated places, so that they could not imitate each other. The genetic nature of the "Uner Tan Syndrome" is consistent with the punctuated evolution. The results are consistent with the single gene theory, suggesting a single gene controlling multiple behavioral traits, and the psychomotor theory, suggesting a co-evolution of the human mind, an emergent property of the motor system expressed by human language. It was suggested that a mutation in a single gene responsible for the evolution of the extensor motor system creating "Homo

Erectus" would be enough for the co-evolution of the emergent human mind, according to psychomotor theory (see Tan, 2005a).

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