

Congenital Cranio-Cervical Compression and Metabolic Failure : A Case Report on the "Somatic Triad"

Pathophysiology of a Congenital Neuro-Metabolic & Structural Disorder

Primary Investigator: Crystal Martinez

Date: December 7, 2025

Classification: Rare Genetic/Metabolic Disease (Proposed ICD Entry)

1. ABSTRACT

This Clinical Case Report establishes the definition of Martinez Syndrome, a novel congenital neuro-metabolic disorder historically misdiagnosed under the psychiatric umbrella of "Autism Spectrum Disorder" (specifically Kanner-type/Profound Autism). The etiology is identified as Gametic Epigenetic Asymmetry leading to Rigid DNA Expression and a systemic metabolic crash at approximately 10 weeks gestation. The observed behaviors in this phenotype are reclassified not as psychiatric deficits, but as Manual Autonomic Overrides required to maintain homeostasis due to underlying Cranial-Cervical Structural Failure and Enzymatic Toxicity. This reclassification effectively dissolves the "Autism" label for this specific population, replacing it with a quantifiable and treatable medical diagnosis.

2. ETIOLOGY: THE "10-WEEK CRASH"

The origin of Martinez Syndrome is identified as a critical mismatch in parental methylation at fertilization (**Gametic Epigenetic Asymmetry**). This mismatch creates a "Rigid DNA" expression profile that fails to execute complex developmental instructions.

- **Mechanism: Translational Epigenetic Dysregulation.** The rigid expression profile prevents the necessary plasticity during early fetal development.
- **The Event:** A metabolic and structural stall occurs at the **10-Week Gestational Window** (the timing of metopic suture formation).
- **The Outcome:** The body prioritizes vital organ development over structural flexibility, resulting in **Metopic Synostosis** and **C1 Subluxation**.

3. CLINICAL PRESENTATION: THE MARTINEZ BIOMARKERS

A. Structural Biomarkers (The Hardware Failure)

The patient presents with extensive skeletal remodeling consistent with chronic manual compensation for congenital instability.

Figure 1: Metopic Synostosis (The Ridge)

Physical examination confirms a prominent vertical ridge on the forehead, indicating premature fusion of the frontal sutures. This fusion restricts frontal lobe expansion and necessitates the "Frontal Bossing" phenotype.



Figure 1. Clinical Presentation of Metopic Synostosis.

Figure 2: C1 Atlas Subluxation (The Block)

Radiographic imaging confirms a rotational blockage ($>3^\circ$) at the brainstem level. This "kink" compresses the vagus nerve and restricts Cerebrospinal Fluid (CSF) drainage, creating a "Hydraulic Lock."

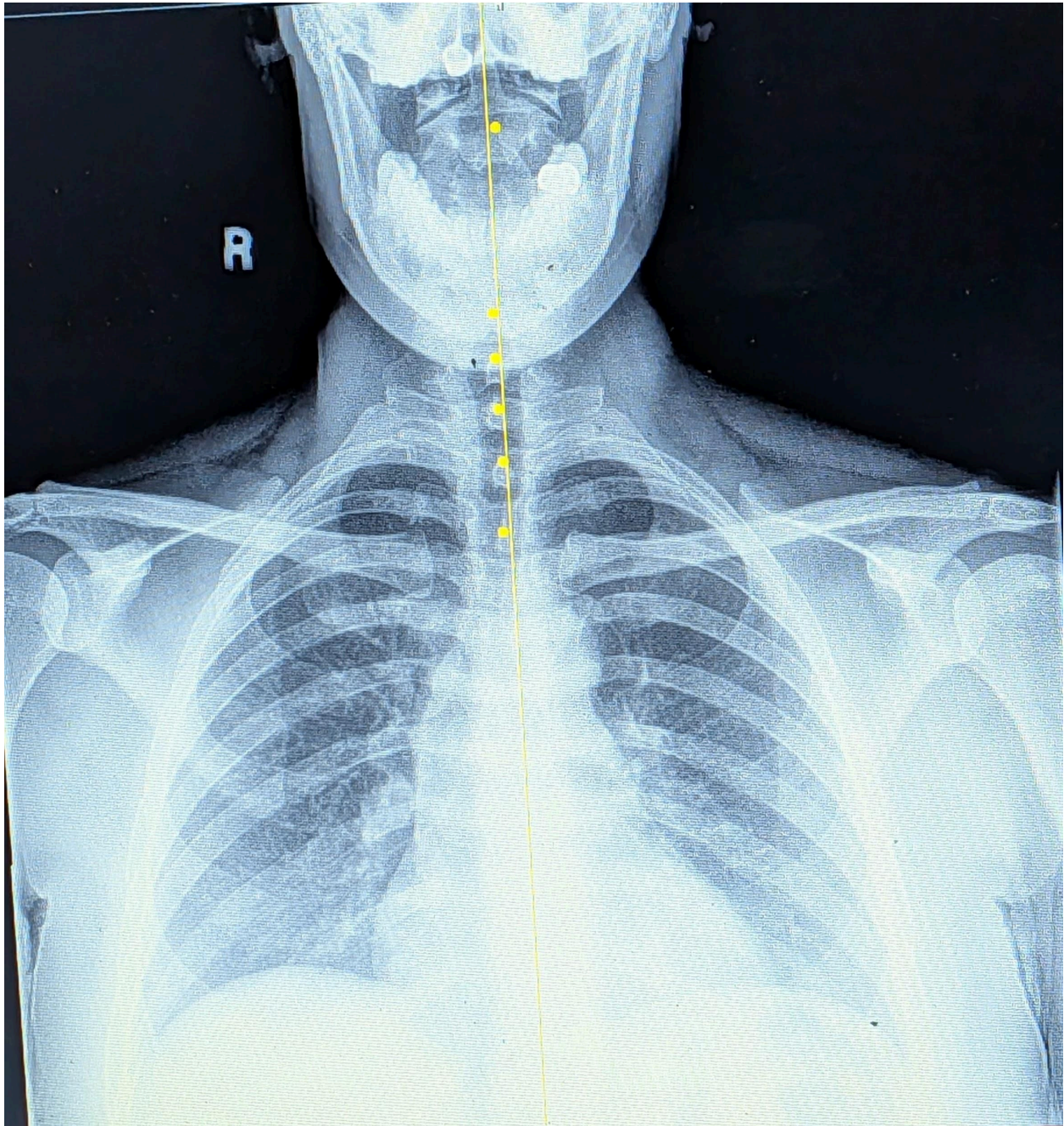


Figure 2. Rotational Subluxation of the Atlas (C1).

Figure 3: Spinal Compensation (Bone Spurs) Thoracic imaging reveals **Anterior Osteophytes** (Bone Spurs) and loss of cervical lordosis. These calcifications are forensic evidence of chronic structural stress required to maintain spinal integrity against the C1 failure.

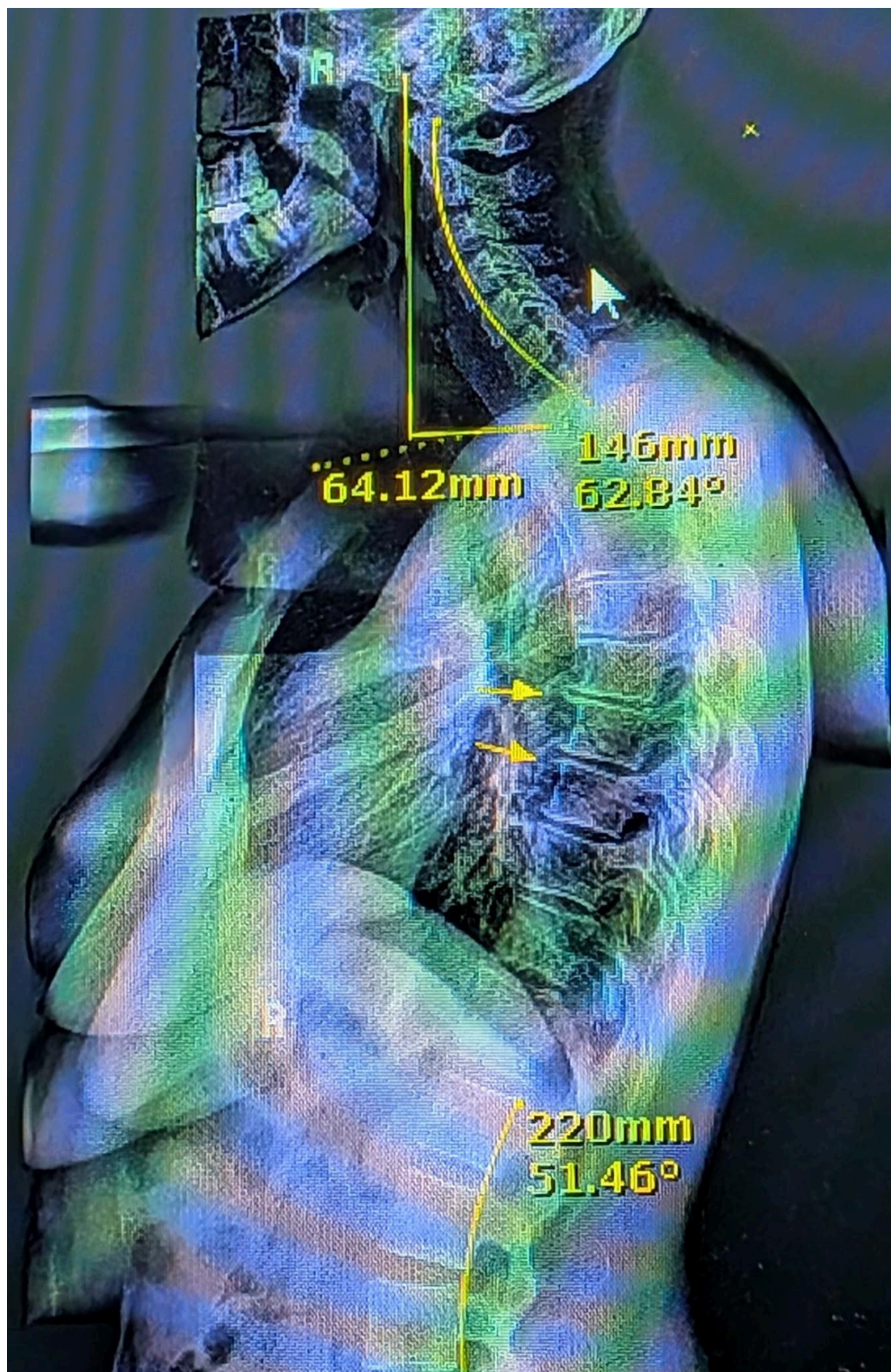


Figure 3. Chronic Spinal Compensation (Anterior Osteophytes).

B. Metabolic Biomarkers (The Toxicity & Velocity Trap)

Pharmacogenomic analysis confirms the enzymatic failure responsible for the patient's history of "drug intolerance," neuroleptic toxicity, and coma.

Figure 4: Pharmacogenomic Profile

- **Glucuronidation Failure: UGT1A1*28/*28** (Gilbert's Syndrome). The liver lacks the capacity to clear bilirubin or antipsychotics, leading to neuroleptic toxicity.
- **Clearance Blockade: CYP3A5*3/*3** (Poor Metabolizer).
- **Ultrarapid Metabolic Velocity: CYP1A2*30/*30** (Ultrarapid Metabolizer). This correlates with rapid gastric emptying (~30 mins) and requires specific bio-available llgfdietary density every 4 hours to prevent catabolic crash.

Figure 4. Pharmacogenomic Metabolic Profile.

Medication Response Genetic Test

Patient/Client:
Crystal Martinez

DOB: Jun 25, 1988
ID: 31285500052
Sex assigned at birth: Female

Specimen: Saliva
Barcode: L3662798
Report date: Aug 27, 2025
Specimen collected: Aug 6, 2025
Specimen received: Aug 14, 2025

Ordering provider:
Avantika Varma MD
NPI: 1760825905

CYP3A5 gene

Poor metabolizer

Your poor metabolizer result in the *CYP3A5* gene does not typically impact decisions about medications. Therefore no action is currently needed. This is a common result.

TECHNICAL DETAILS
Diplotype: *3/*3

CYP4F2 gene

Intermediate metabolizer

 Your result in this gene may impact decisions about certain medications.

TECHNICAL DETAILS
Diplotype: *1/*3
Variant detected: rs2108622 CT

DPYD gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS
Diplotype: reference/reference
Activity score: 2.0

NUDT15 gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS
Diplotype: *1/*1

TPMT gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS
Diplotype: *1/*1

UGT1A1 gene

Poor metabolizer

 Your result in this gene may impact decisions about certain medications. Some medications could cause serious side effects.

TECHNICAL DETAILS
Diplotype: *28/*28

color

Color Diagnostics LLC
863 Mitten Road, Burlingame, CA 94010
CLIA #05D2081492 - CAP #8975161 - NY #9069

(844) 352-6567
support@color.com

Lab Director CLIA: Dr. Russell Kerschmann, MD
Lab Director NY: Dr. Scott Topper, PhD, FACMG

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Metabolizer genes

CYP1A2 gene

Ultrarapid metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: *30/*30

CYP2C cluster

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: reference/reference

CYP2C19 gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: *1/*1

CYP2C9 gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: *1/*1
Activity score: 2.0

CYP2D6 gene

Normal metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: *1/*1
Activity score: 2.0

CYP3A4 gene

Intermediate metabolizer

Your result in this gene does not typically impact decisions about medications. Therefore, no action is currently needed.

TECHNICAL DETAILS

Diplotype: *1/*22

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Figure 4. Pharmacogenomic Metabolic Profile.

C. Neuropsychological Profile (The "Spiky" 2E)

The patient presents with a distinct **Twice-Exceptional (2E)** profile: Superior Intelligence obscured by profound somatic disability.

Figure 5: Longitudinal Evidence of Superior Intellectual Function

School records verify **93rd Percentile** performance in Spelling and **85th Percentile** in Language Mechanics, occurring simultaneously with documentation of "ungovernable" behavioral failures. This discrepancy confirms that the pathology is not cognitive, but a **Metabolic/Structural Interference** preventing consistent output.

TEST RESULTS

NAME

GRADE

SSN

DOB

TEST DATE

TEST TIME

TEST TYPE

TEST LOCATION

TENNESSEE COMPREHENSIVE ASSESSMENT PROGRAM

APRIL, 1996 SCORES

CTRS/4 SCORES	READING VOC COMP	LANGUAGE MECH EXPR	MATHEMATICS C&A TOTL	TOTL WORD BATT ONLY	SCI SPEL	SDC STD
SS	624	658	641	669	695	680
NCE	42	50	46	58	51	63
RS	4	5	3	6	6	6
NP	36	51	43	65	61	73
RANGE	23-36	32-51	52-65	43-65	51-61	58-73
GRADE: 2.7	53	60	80	76	81	85
LEVEL: 12						
SURVEY FORM: C						
NORMS FROM: 1988						
CRD. CODES						

TENNESSEE DOMAIN SCORES

MECHANICS	H	SER/PARA	H	SPL/MD ID	H	COMPREH	H
REF STUDY	H	NUMERATH	H	WHOLE/INT	H	FRACTIONS	H
GRAPHS	H	GEOMETRY	H	PROB SOLV	H		

TENNESSEE COMPREHENSIVE ASSESSMENT PROGRAM

APRIL, 1997 SCORES

CTRS/4 SCORES	READING VOC COMP	LANGUAGE MECH EXPR	MATHEMATICS C&A TOTL	TOTL WORD BATT ONLY	SCI SPEL	SDC STD
SS	680	717	699	666	694	680
NCE	52	64	59	40	45	47
RS	5	6	6	4	5	4
NP	54	75	67	32	41	36
RANGE	38-60	54-75	20-32	26-41	26-41	26-41
GRADE: 3.7	71	87	78	48	58	48
LEVEL: 13						
SURVEY FORM: H						
NORMS FROM: 1988						
CRD. CODES						

TENNESSEE DOMAIN SCORES

MECHANICS	H	LANG USE	H	SER/PARA	H	SPL/MD ID	H	COMPREH	P	DECIMALS	H
REF STUDY	H			NUMERATH	H	WHOLE/INT	H	FRACTIONS	P		
GRAPHS	H	MEASUREMENT	P	GEOMETRY	H	PROB SOLV	P				

TCAP - WRITING ASSESSMENT

02/05/98

SCORE
3.0

TCAP M	READING	LANGUAGE	MATHEMATICS	TOTL	SCI	SDC
SS	624	658	641	669	695	680
NCE	42	50	46	58	51	63
RS	4	5	3	6	6	6
NP	36	51	43	65	61	73
RANGE	23-36	32-51	52-65	43-65	51-61	58-73
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LEVEL: 12						
SURVEY FORM: C						
NORMS FROM: 1988						
CRD. CODES						

TCAP M
READING
LANGUAGE
MATHEMATICS
TOTL
SCI
SDC

SS
NCE
RS
NP
RANGE
GRADE
LEVEL
SURVEY FORM
NORMS FROM
CRD. CODES

02/05/98
04/02
DATE
GRADE
M-17
NORMS DATE: 1996
PATTERN MONTH: 31
PATTERN (IRT)
M009634001-04-15755

Figure 5. Longitudinal Evidence of Superior Intellectual Function (The 2E Profile).

MEMBER
11th Grade Female (Form 23F)
SSN: XXX XX-XX-XXXX
Test Date: Nov 2, 2005
High School

Form No 0021

ASVAB SUMMARY RESULTS

ASVAB Results

ASVAB Results	Percentile Scores		11th Grade Standard Score Bands		11th Grade Standard Scores
	11th Grade Female	11th Grade Males	11th Grade Students		
Career Exploration Scores	81	80	81		58
Verbal Skills	63	64	63		53
Math Skills	81	61	71		54
Science and Technical Skills					
ASVAB Tests					
General Science	70	61	66		54
Arithmetic Reasoning	64	60	62		52
Word Knowledge	69	66	67		54
Paragraph Comprehension	93	95	94		63
Mathematics Knowledge	59	64	61		53
Electronics Information	72	47	60		52
Auto and Shop Information	45	20	32		44
Mechanical Comprehension	89	69	79		57
Military Entrance Score (AFQT) 60					

EXPLANATION OF YOUR ASVAB PERCENTILE SCORES

Your ASVAB results are reported as percentile scores in the three highlighted columns to the left of the graph. Percentile scores show how you compare to other students - males and females, and for all students - in your grade. For example, a percentile score of 65 for a 11th grade female would mean she scored better than 65 out of every 100 females in the 11th grade.

For purposes of career planning, knowing your relative standing in these comparison groups is important. Being in the top female does not limit your career or educational choices. There are noticeable differences in how men and women score in some areas. Viewing your scores in light of your relative standing both to men and women may encourage you to explore areas that you might otherwise overlook.

You can use the Career Exploration Scores to evaluate your knowledge and skills in three general areas: Verbal, Math, and Science and Technical Skills. You can use the ASVAB Test Scores to gather information on specific skill areas. Together, these scores provide a snapshot of your current knowledge and skills. This information will help you develop and review your career goals and plans.

EXPLANATION OF YOUR ASVAB STANDARD SCORES

Your ASVAB results are reported as standard scores in the above graph. Your score on each test is identified by the "X" in the corresponding bar graph. You should view these scores as estimates of your true skill level in that area. If you took the test again, you probably would receive a somewhat different score. Many things, such as how you were feeling during testing, contribute to this difference. This difference is shown with gray score bands in the graph of your results. Your standard scores are based on the ASVAB tests and composites based on your grade level.

The score bands provide a way to identify some of your strengths. Overlapping score bands mean your true skill level is similar in both areas, so the real difference between specific scores might not be meaningful. If the score bands do not overlap, you probably are stronger in the area that has the higher score band.

The ASVAB is an aptitude test. It is neither an absolute measure of your skills and abilities nor a perfect predictor of your success or failure. A high score does not guarantee success, and a low score does not guarantee failure. In a future educational program or occupation, for example, if you have never worked with shop equipment or cars, you may not be familiar with the terms and concepts

assessed by the Auto and Shop Information test. Taking a course or obtaining a part-time job in this area would increase your knowledge and improve your score if you were to take it again.

USING ASVAB RESULTS IN CAREER EXPLORATION

Your career and educational plans may change over time as you gain more experience and learn more about your interests. *Exploring Careers: The ASVAB Career Exploration Guide* can help you learn more about yourself and the world of work, to identify and explore potential goals and develop an effective strategy to realize your goals. The Guide will help you identify occupations in line with your interests and skills. As you explore potentially satisfying careers, you will develop your career exploration and planning skills.

Meanwhile, your ASVAB results can help you in making well-informed choices about future high school courses.

We encourage you to discuss your ASVAB results with a teacher, counselor, parent, family member or other interested adult. These individuals can help you review your ASVAB results in light of other important information, such as your interests, school grades, motivation, and personal goals.

USE OF INFORMATION

Personal identity information (name, social security number, street address, and telephone number) and test scores will not be released to any agency outside of the Department of Defense (DoD), the Armed Forces, the Coast Guard, and your school. Your school or local school system can determine any further release of information. The DoD will use your scores for recruiting and research purposes for up to two years. After that, the information will be used by the DoD for research purposes only.

MILITARY ENTRANCE SCORES

The Military Entrance Score (also called AFQT, which stands for the Armed Forces Qualification Test) is the score used to determine your qualifications for entry into any branch of the United States Armed Forces or the Coast Guard. The Military Entrance Score provides a general way how well you might do in training and on the job in military occupations. Your score reflects your standing compared to Americans men and women 18 to 25 years of age.

Use Access Code: 378803804W

(for online Occu-Find and FYI)

Access Code expires: Jul 15, 2007

Explore career possibilities by using your Access Code at
www.asvabprogram.com
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Page

Self-Reported Medical Conditions

Figure 5.1 DSM-4

Psychiatric Misdiagnosis: The patient's metabolic distress was historically labeled as **Oppositional Defiant Disorder (ODD)**, **Panic Syndrome**, and **Severe Anhedonia**. These are reclassified as symptoms of **Metabolic Encephalopathy** and **Autonomic Storms**.

Figure 6: Thermal Neuro-Scanning (The Inflammation)

Paraspinal thermal imaging confirms significant thermal asymmetry at the C1 level, recording a 3.1-degree Fahrenheit differential. This extreme delta indicates acute and chronic neurological interference at the brainstem, corroborating the radiographic findings of rotational subluxation. The thermal spike serves as a biomarker for the localized inflammation driving the "Rerouting" signal failure.

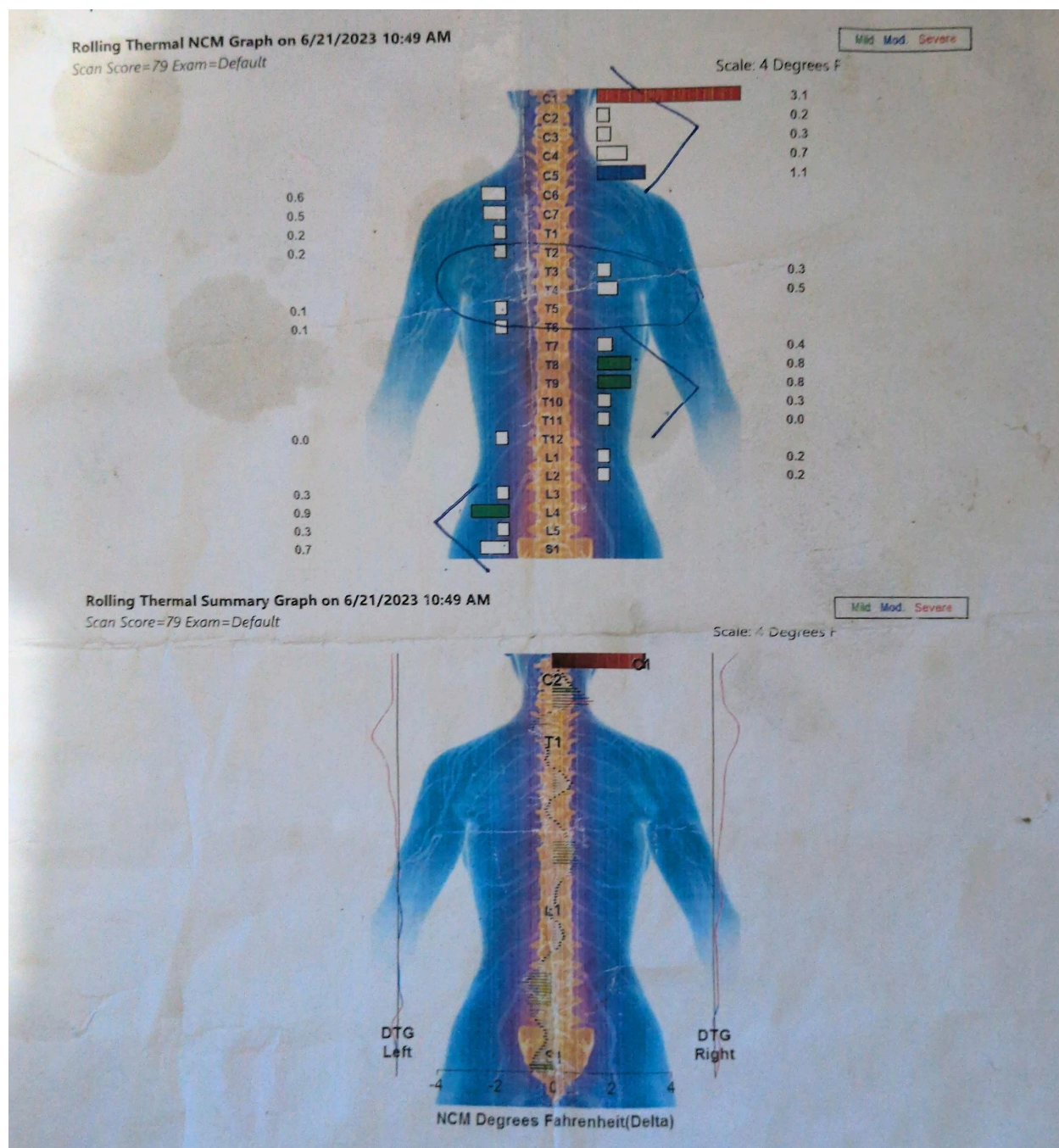


Fig 6. Rolling Thermal Scan verifying 3.1-degree differential at C1.

D. Somatic Biomarkers (Manual Life Support)

Behaviors historically labeled as "stimming" are reclassified as **Mechanical Survival Functions**:

- **The Hydraulic Pump (Head Banging):** Ballistic shockwaves generated by rhythmic impact are utilized to force Cerebrospinal Fluid (CSF) through the restricted C1 valve and relieve cranial pressure caused by the **Metopic Ridge**.
- **The "Second Heart" (Toe Walking):** Due to autonomic vascular failure, the patient utilizes "Toe Walking" to maintain permanent contraction of the gastrocnemius muscles, manually forcing venous blood return to the brain against gravity to prevent syncope.
- **Reverse Swallow & Acid Reflux:** Persistent "Reverse Swallow" pattern (tongue thrust) accompanied by chronic Acid Reflux indicates congenital failure of the glossopharyngeal nerve mechanism. The patient must manually regulate swallowing to prevent aspiration.
- **Social Oblivion:** A state of cortical unavailability due to the allocation of all processing power to these autonomic regulation tasks.

4. PATHOPHYSIOLOGY: THE MANUAL SYSTEM RESET

In Martinez Syndrome, the autonomic nervous system fails to regulate basic survival functions automatically.

- **The Cortisol Mechanism:** The body perceives the structural blockage and metabolic failure as a constant "Internal Emergency," leading to chronic HPA Axis activation and elevated cortisol (The "Alarm").
- **The Rerouting:** Because normal pathways are blocked (Inflammation/C1), the body attempts to "reroute" signals.
- **The Manual Reset:** When the system glitches or locks up, the patient utilizes high-impact somatic behaviors (Head Banging) as a **"Manual System Reset"** to force the neurological grid to reboot and restore flow.

5. ETHICAL IMPLICATIONS: THE CHEMICAL LOBOTOMY

The administration of antipsychotics to patients with Martinez Syndrome constitutes **Iatrogenic Negligence**.

- **Mechanism:** Due to **UGT1A1 Failure**, these agents bio-accumulate into neurotoxins.
- **Impact:** These agents suppress the frontal cortex—the patient's primary **Manual Override** for survival.
- **Case Evidence:** The subject suffered a **2-Week Coma** and vital collapse following standard psychiatric intervention, confirming the lethal incompatibility of these agents with the Martinez phenotype.

6. CONCLUSION: THE END OF THE "AUTISM" LABEL

This case report demonstrates that the phenotype historically known as "Profound Autism" (in this subset) is, in fact, **Martinez Syndrome**: a quantifiable **Neurometabolic and Structural Disease**.

- **The "Autism" Diagnosis is Rejected:** The behavioral symptoms are secondary effects of structural compression and metabolic toxicity.
- **The "Asperger's" Distinction:** This phenotype is biologically distinct from the **Mimetic Personality Adaptation** (formerly Asperger's), which is an environmental trauma response.
- **The New Standard:** Patients presenting with the **Martinez Triad** (Ridge, C1, UGT1A1) must be removed from the psychiatric "spectrum" and treated with **Orthopedic Decompression** and **Pharmacogenomic-Guided Metabolic Support**. "Autism" is no longer a valid diagnosis for this medical condition.