AVALIAÇÃO DE HABILIDADES COGNITIVAS E PREFERÊNCIAS SOCIAIS NA SÍNDROME DE RETT

European RETT SYNDROME Conference Maastricht The Netherlands

ABRE-TE Associação Brasileira da Síndrome
São Paulo - Brasil

Universidade Presbiteriana Mackenzie

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Pacientes com síndrome de Rett (RS) apresentam severos prejuízos motores, de linguagem e cognitivos, bem como perda dos movimentos manuais espontâneos. Por outro lado, há robustas evidências de que estas pessoas utilizam os movimentos oculares de forma intencional.

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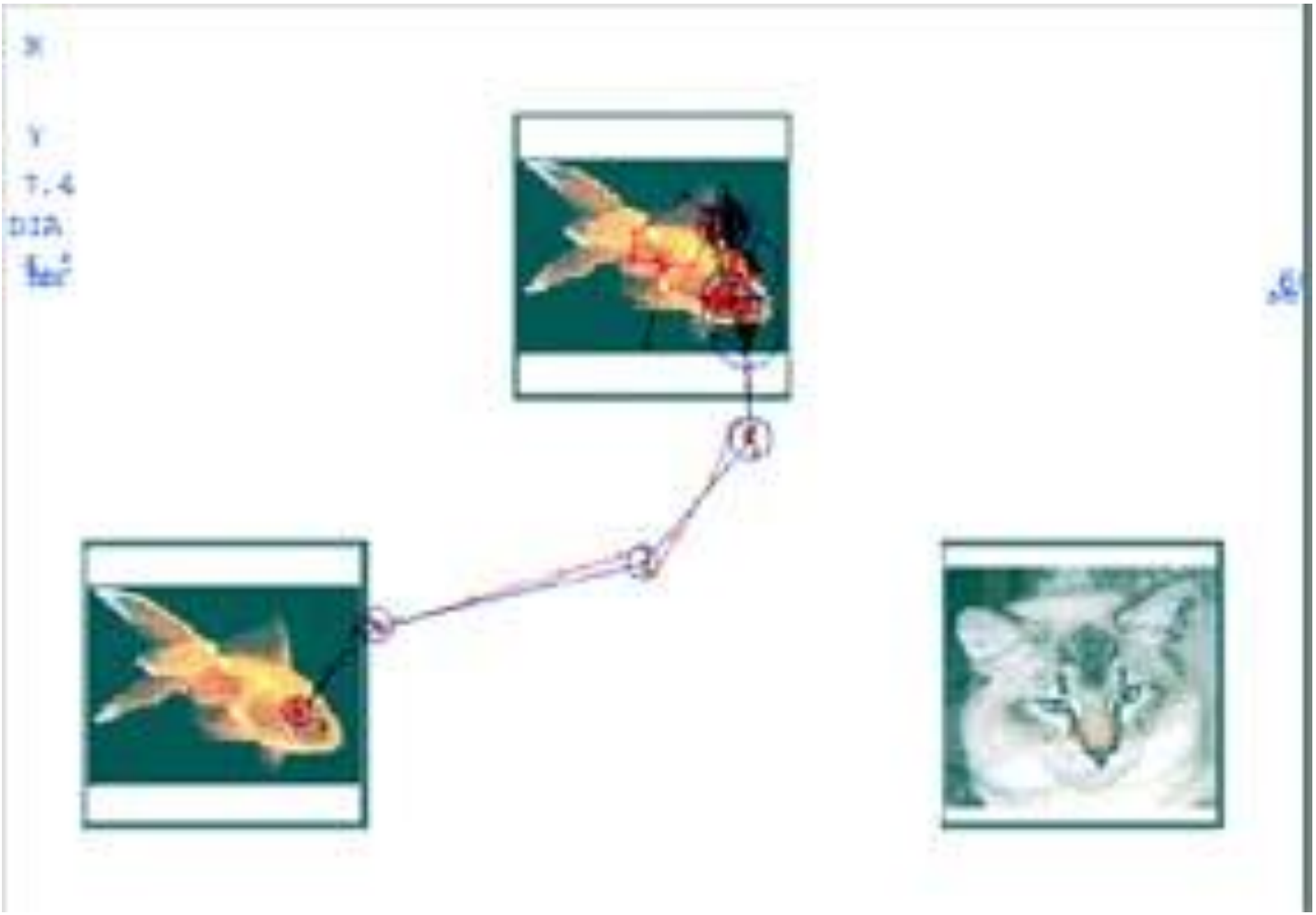
Cognitive performance in Rett syndrome girls: a pilot study using eyetracking technology

P. M. Baptista, M. T. Mercadante, E. C. Macedo & J. S. Schwartzman

Development Disorders Program, Mackenzie Presbyterian University, São Paulo, Brazil

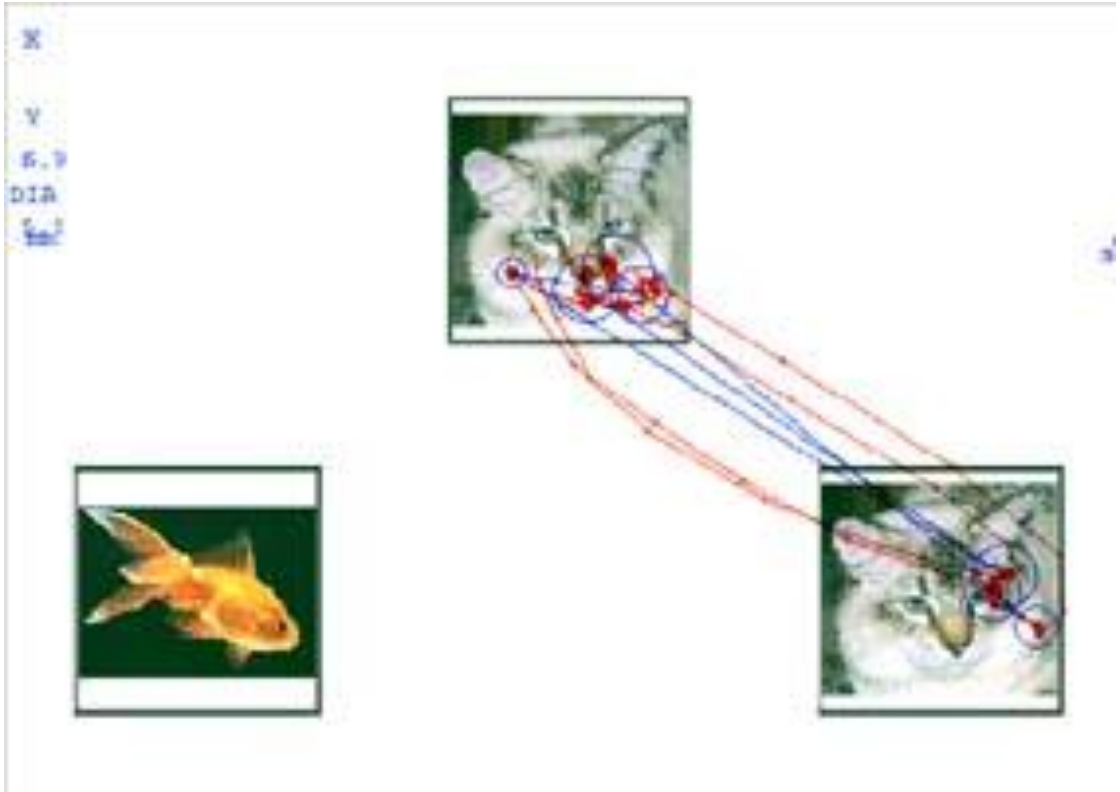
Pudemos demonstrar que o rastreamento visual pode ser utilizado em Pessoas com SR e que estas podem entender e olhar para determinada figuras solicitadas.

Rett 4 anos: olhe o peixinho

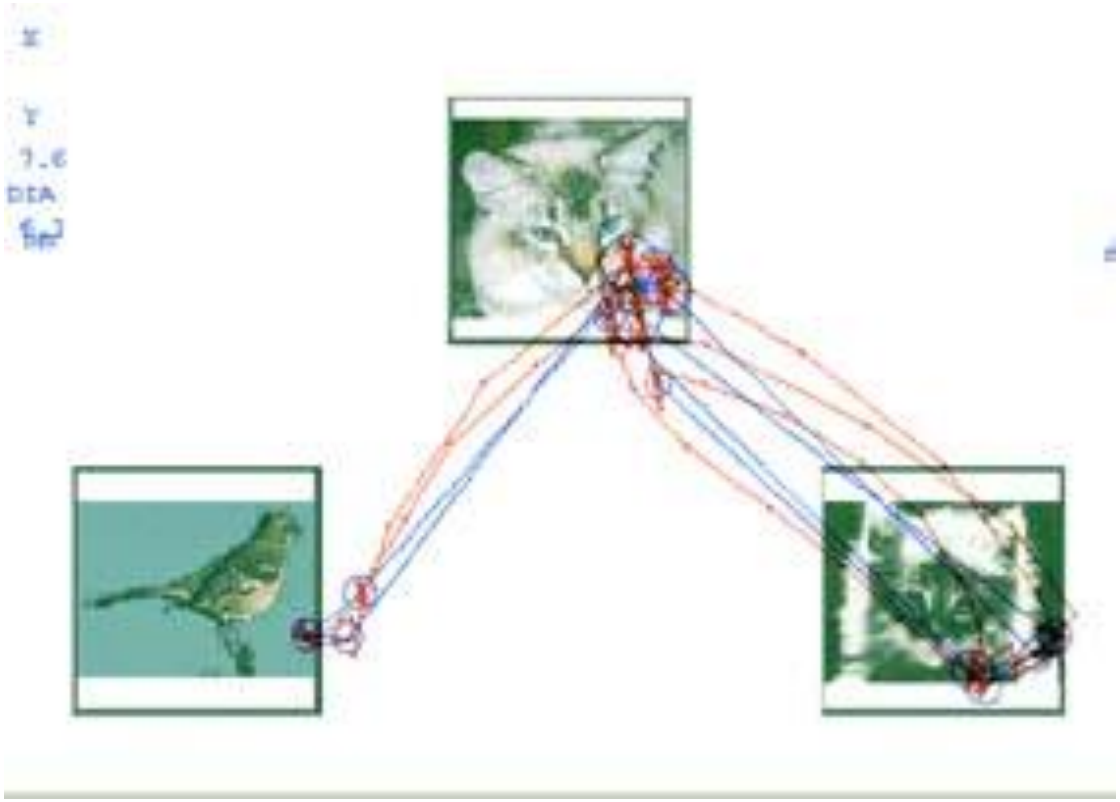


Rett 4 anos: onde está o outro peixinho?

9 anos de idade: olhe para o gatinho



onde está o outro gatinho igual?



onde está o outro gatinho diferente?

Arq Neuropsiquiatr 2009;67(1):50-54

CONCEPTS OF COLOR, SHAPE, SIZE AND POSITION IN TEN CHILDREN WITH RETT SYNDROME

Renata de Lima Velloso[1], Ceres Alves de Araújo[2], José Salomão Schwartzman[1]

Abstract – Individuals with Rett syndrome (RS) present severe motor, language and cognitive deficits, as well as spontaneous hand movement loss. On the other hand, there are strong evidence that these individuals use the eyes with intentional purpose. Ten girls aged 4y8m to 12y10m with RS were assessed with a computer system for visual tracking regarding their ability of indicating with eyes the recognition of concepts of color (red, yellow and blue), shape (circle, square and triangle), size (big and small) and spatial position (over and under) to which they were first exposed to. Results from comparing the time of eyes fixation on required and not required concepts did not differ significantly. Children did not show with eyes the recognition of the required concepts when assessed with eye tracking system.

KEY WORDS: Rett syndrome, cognition, eye movement measurements, eye movements.

Não encontramos evidências de que as pacientes identificaram os estímulos apresentados. Neste estudo o tempo de exposição prévia aos estímulos não foi controlado e cada slide foi mostrado por 3 segundos.

Conceitos de cor, forma, tamanho e posição e evidências de preferências sociais em 14 pacientes com síndrome de Rett

José Salomão Schwartzman
Silvana Santos
Renata Lima Velloso

Método

- **14 pacientes com Rett syndrome patients**
  idade: de4 a30 anos
- expostas aos estimulos estudados de14 a 45 dias (15 a 30 minutos por dia)

  cor: vermelho, amarelo e azul,
  tamanho: grande e pequeno
  posição: em cima e embaixo

- **meninas típicas**
  15
  idade: de 3 a 6 anos

Método

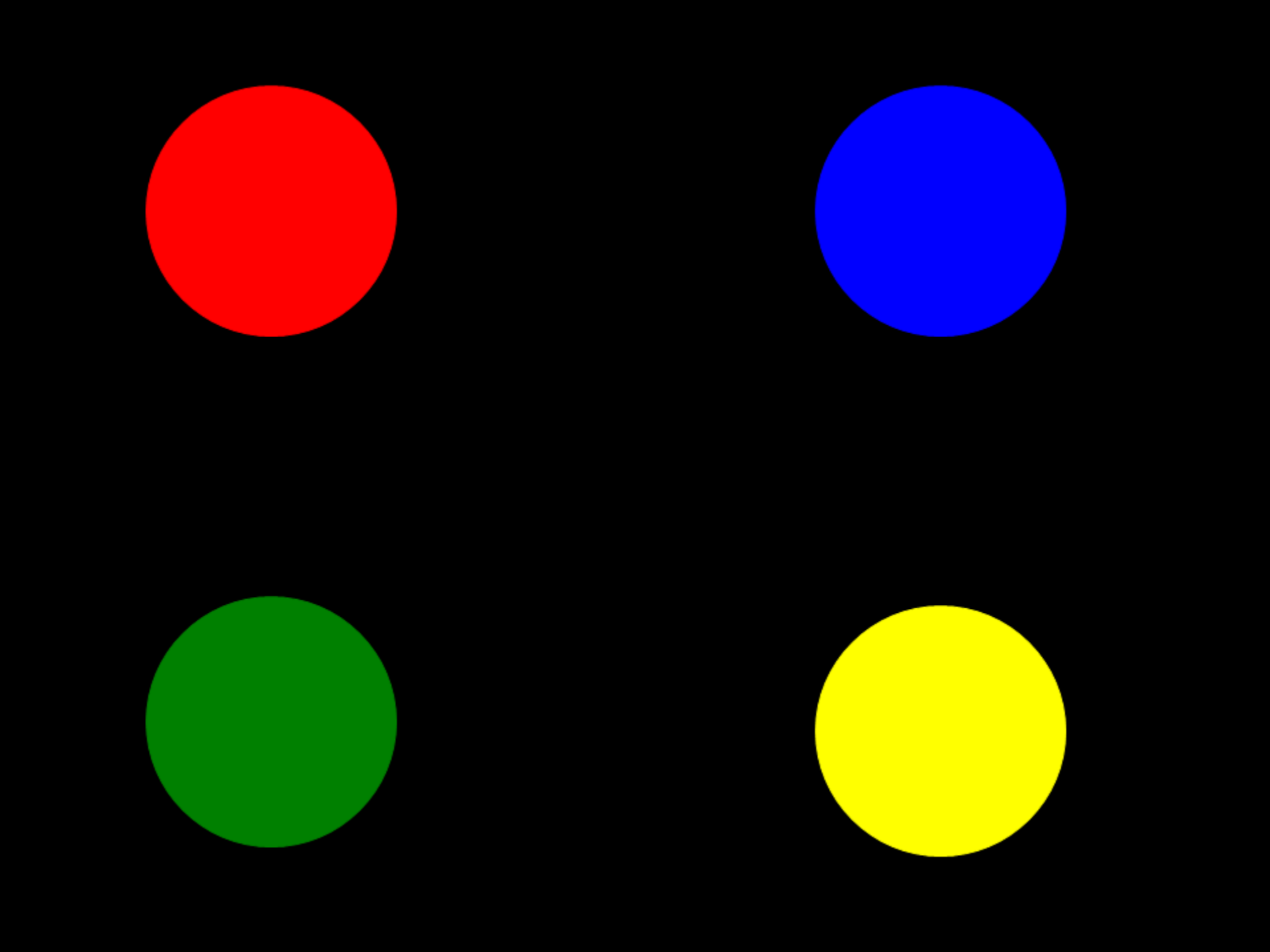
- **Pacientes com SR :** Laboratório de Pesquisas do Programa de Pós-Graduação em Distúrbios do Desenvolvimento da Universidade Presbiteriana Mackenzie utilizando o equipameto computadorizado Tobii 1750 e o software ClearView (Tobii Technology, 2005)

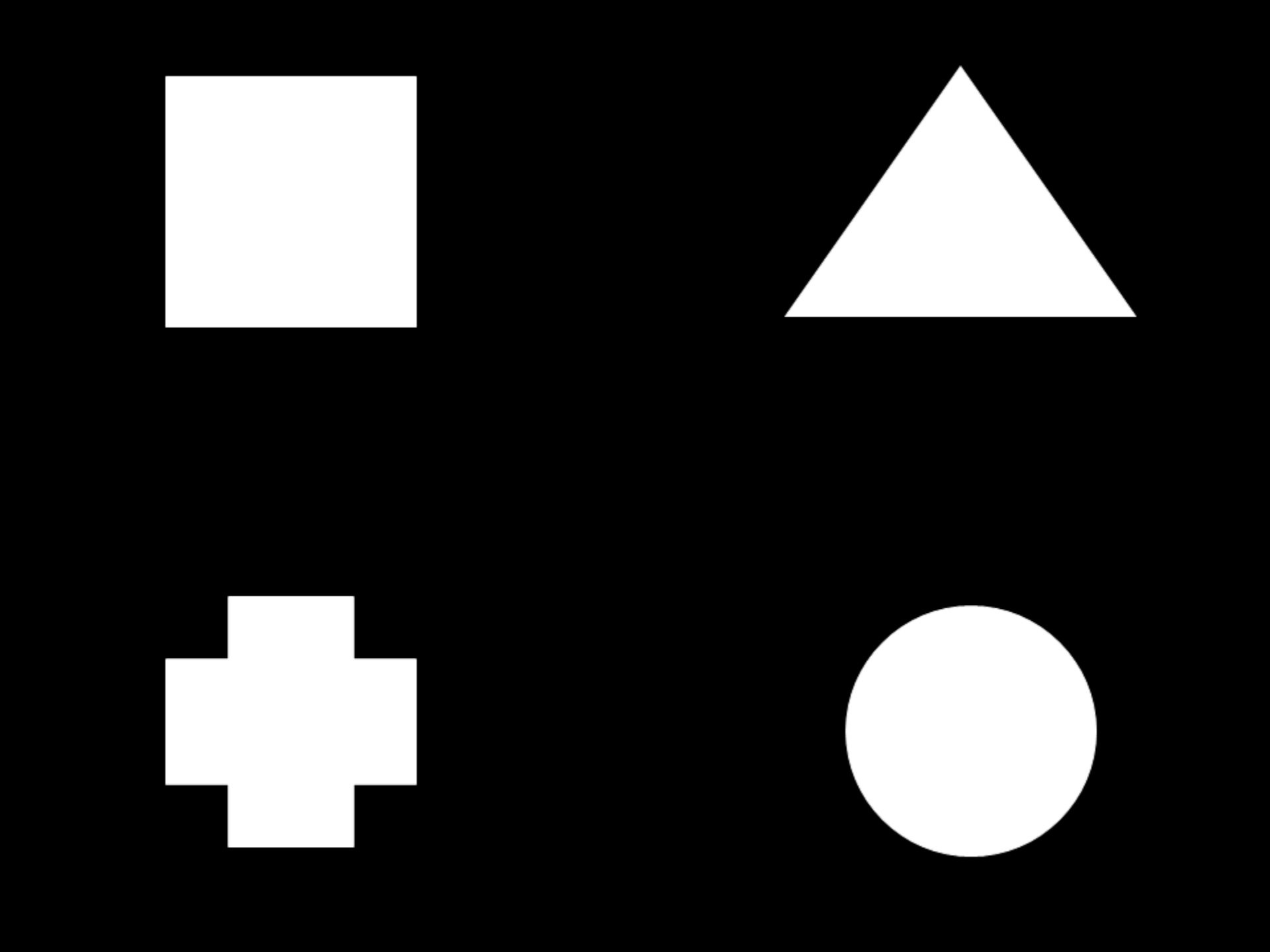
- **Meninas tipicas :** escola privada regular utilizando o eye tracker S2 Mirametrix.

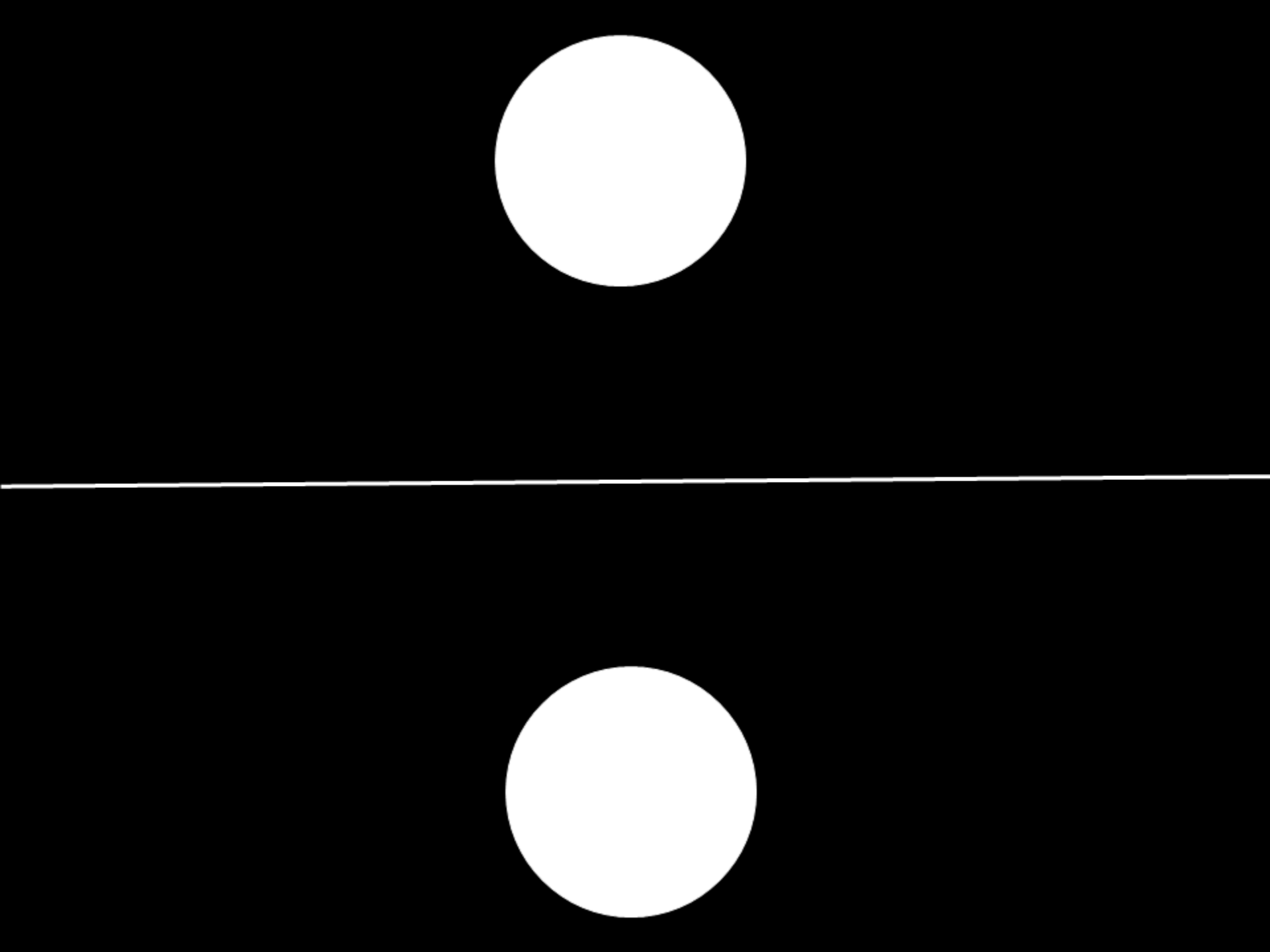
Method

- The first step was the required calibration procedure.
- Then the subjects should look at the computer screen and the instruction was LOOK AT …….(red, bigger, above etc)
- Each picture was presented for 8 seconds, repeated for 3 times with a 3 seconds interval (neutral picture)

  between the presentations







RESULTADOS GRUPO RETT

Caso	Mutação	Right answers				
		Cores (3)	Formas (3)	Tamanho (2)	Posição (2)	Total (10)
1	C236dupC	2	0	0	1	3
2	Não sabe	1	0	0	0	1
3	R306C	2	0	1	0	3
4	P152R	0	1	1	0	2
5	T158M	0	0	0	0	0
6	R106W	2	0	0	1	3
7	negativo	0	0	0	0	0
8	R294X	2	3	1	2	8
9	R106W	0	0	0	0	0
10	Não conclusivo	0	0	1	0	1
11	NI	0	1	0	0	1
12	Negativo	0	0	1	0	1
13	Não realizado	0	0	0	0	0
14	C.1164 del 11	0	0	0	0	0
		9/42	5/42	5/28	4/28	23/140
		21,43%	11,90%	17,86%	14,29%	16,43%

RESULTADOS GRUPO RETT

CasO	Mutação	Respostas corretas				
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		21,43%	11,90%	17,86%	14,29%	16,43%

Respostas corretas X Variáveis

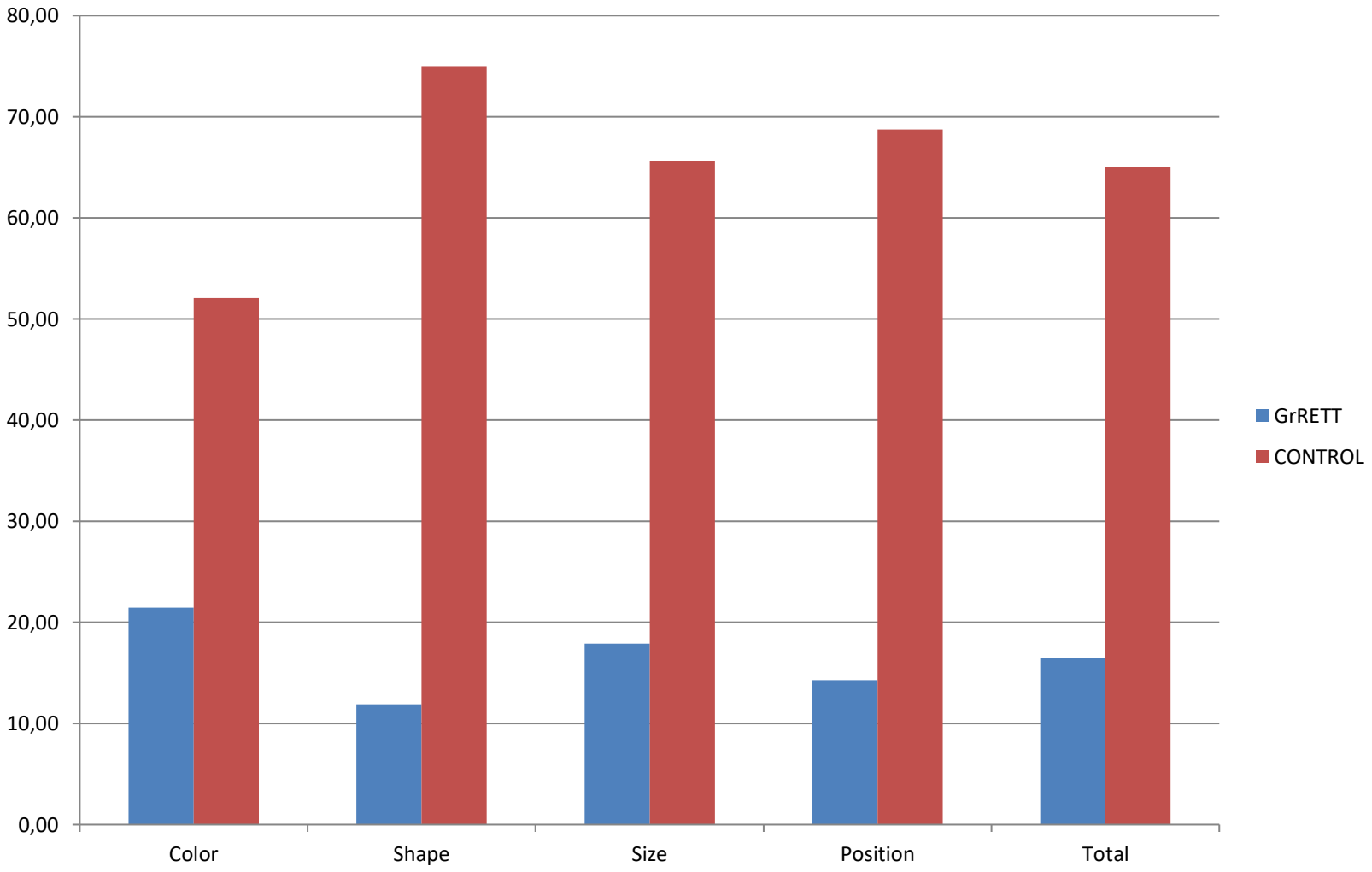
(excluindo caso 8)

Idade	Exposição	Marcha	Estereotipias	Convulsões	Escola
-0.20	-0.15	0.16	0.27	0.24	0.22

(incluindo caso 8)

Idade	Exposição	Marcha	Estereotipias	Convulsões	Escola
0.01	0.17	-0.03	0.00	0.01	-0.10

	Cores	Formas	Tamanho	Posição	Total
grRETT	21.43%	11.90%	17.86%	14.29%	16.43%
CONTROLES	52.08%%	75.00%	65.63%	68.75%	65.00%



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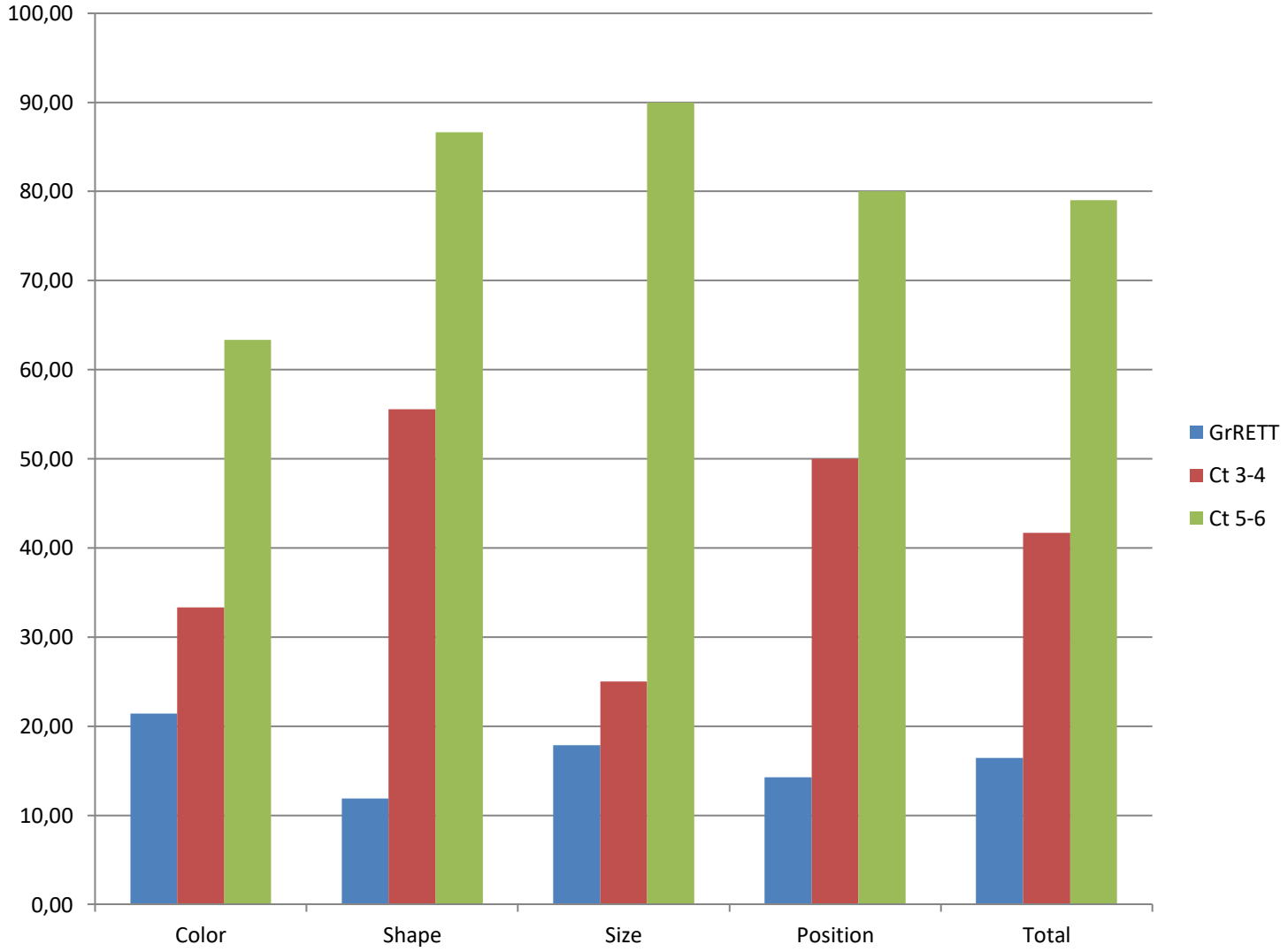


Fourteen girls aged 4 years to 30 years with RS were assessed with a computer system for visual tracking regarding their ability of indicating with eyes the recognition of concepts of color (red, yellow and blue), shape (circle, square and triangle), size (big and small) and spatial position (over and under) to which they were intensively first exposed to.

A comparação do tempo de fixação ocular nos estimulos solicitados e nos não solicitados não mostraram diferenças significativas. As pacientes conseguiram identificar 23 dos 140 estímulos solicitados.

CONTATO VISUALE (Preferências sociais) síndrome de Rett – TEA– Meninas típicas

- 14 pacientes com SR
  - 4 a30 anos
- 11 pacientes TEA
  - Uma menina
  - 4 a20 anos
- 14 meninas tipicas
  - 3 a6 anos

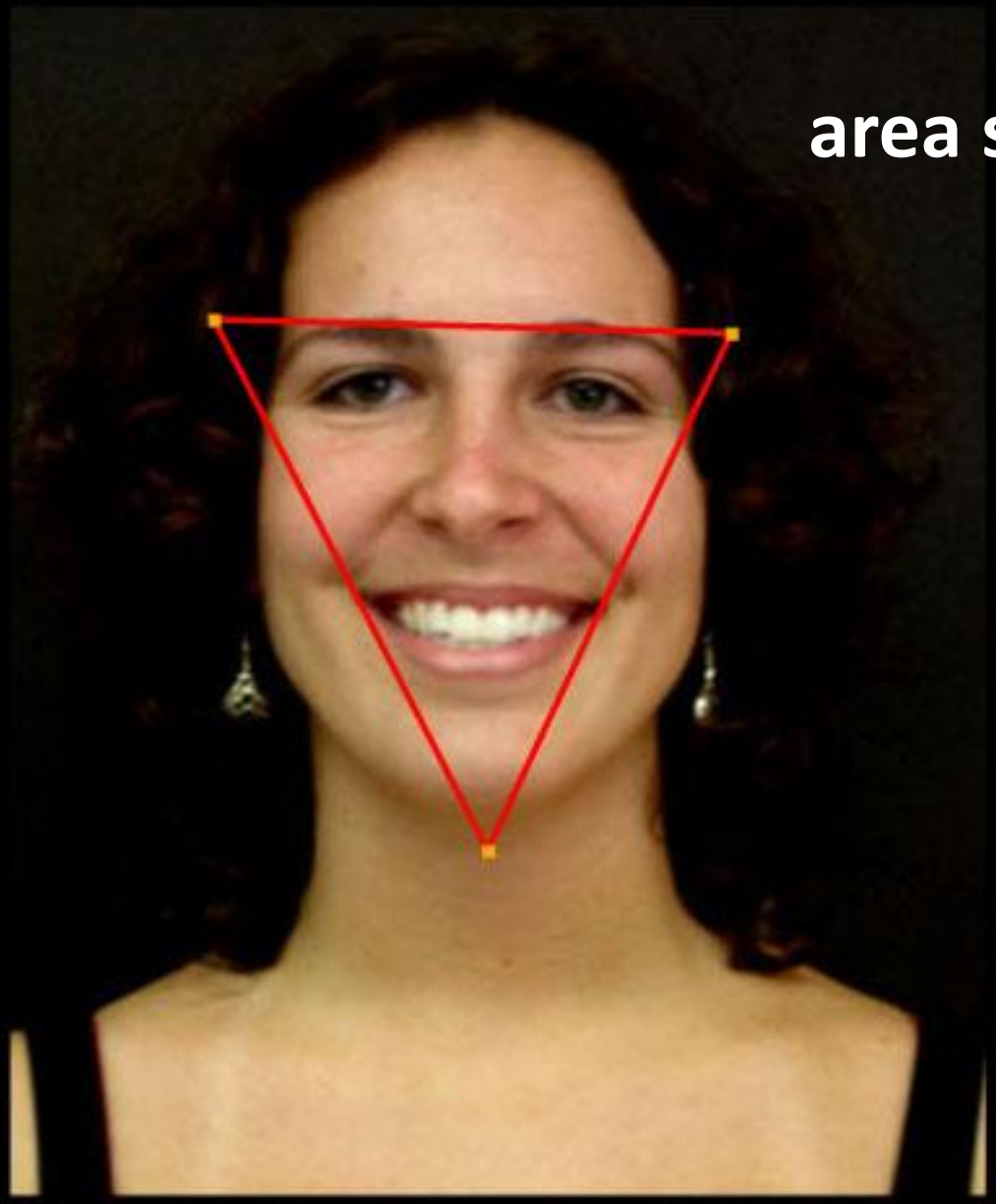
GRUPO TEA

	SUJEITO	SEXO	IDADE/ANOS
1	DM	M	4
2	NL	M	4
3	AC	M	6
4	F	M	6
5	DL	M	6
6	SS	M	7
7	FG	M	8
8	GB	M	10
9	Eu	M	10
10	LN	F	12
11	RM	M	20

CONTATO VISUAL (Preferências sociais) síndrome de Rett – TEA– meninas típicas

- Deveriam olhar para o computador sem nenhuma instrução especial
- Cada figura foi exposta por 8 segundos com intervalos de 3 segundos.
- Não houve exposição prévia às figuras.

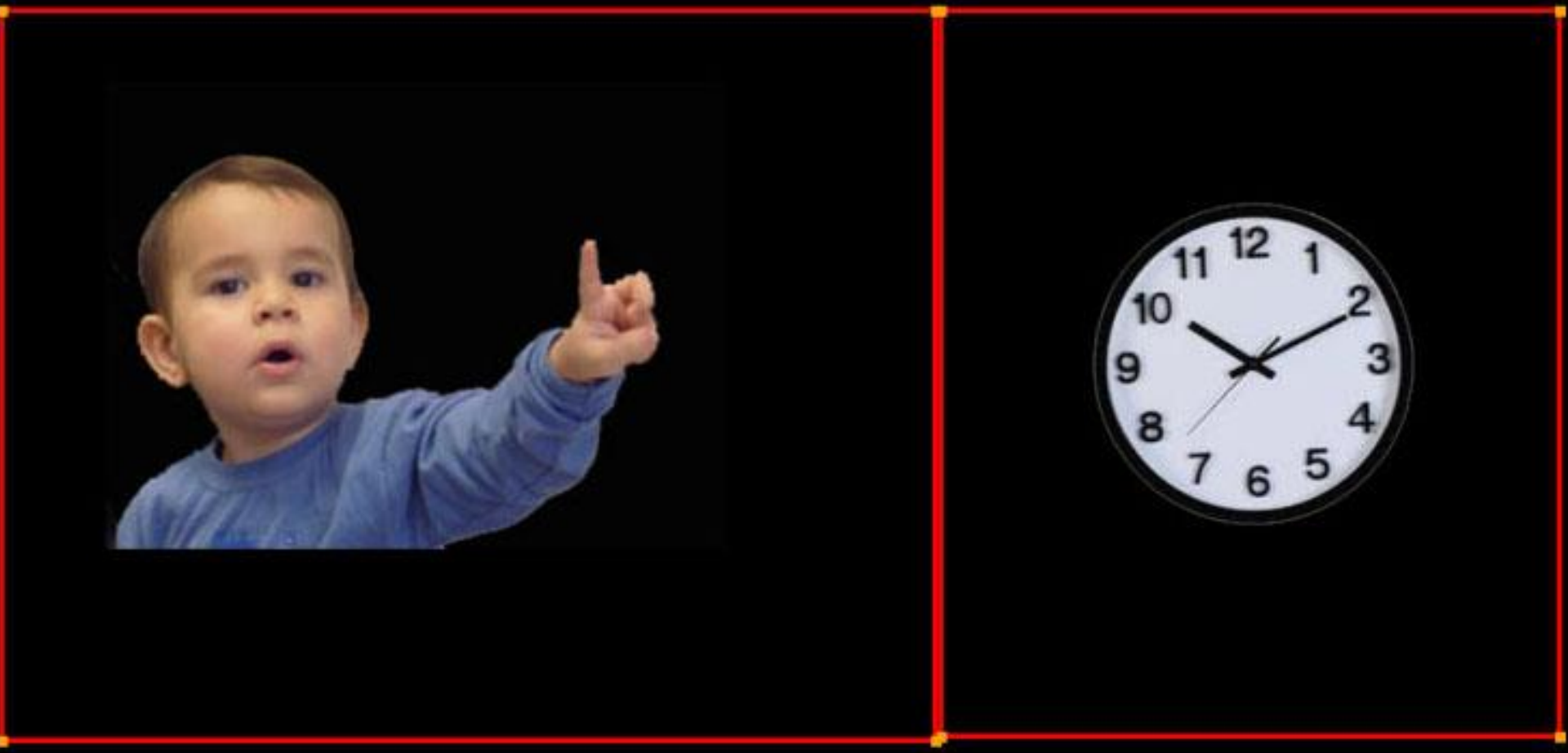
area selecionada

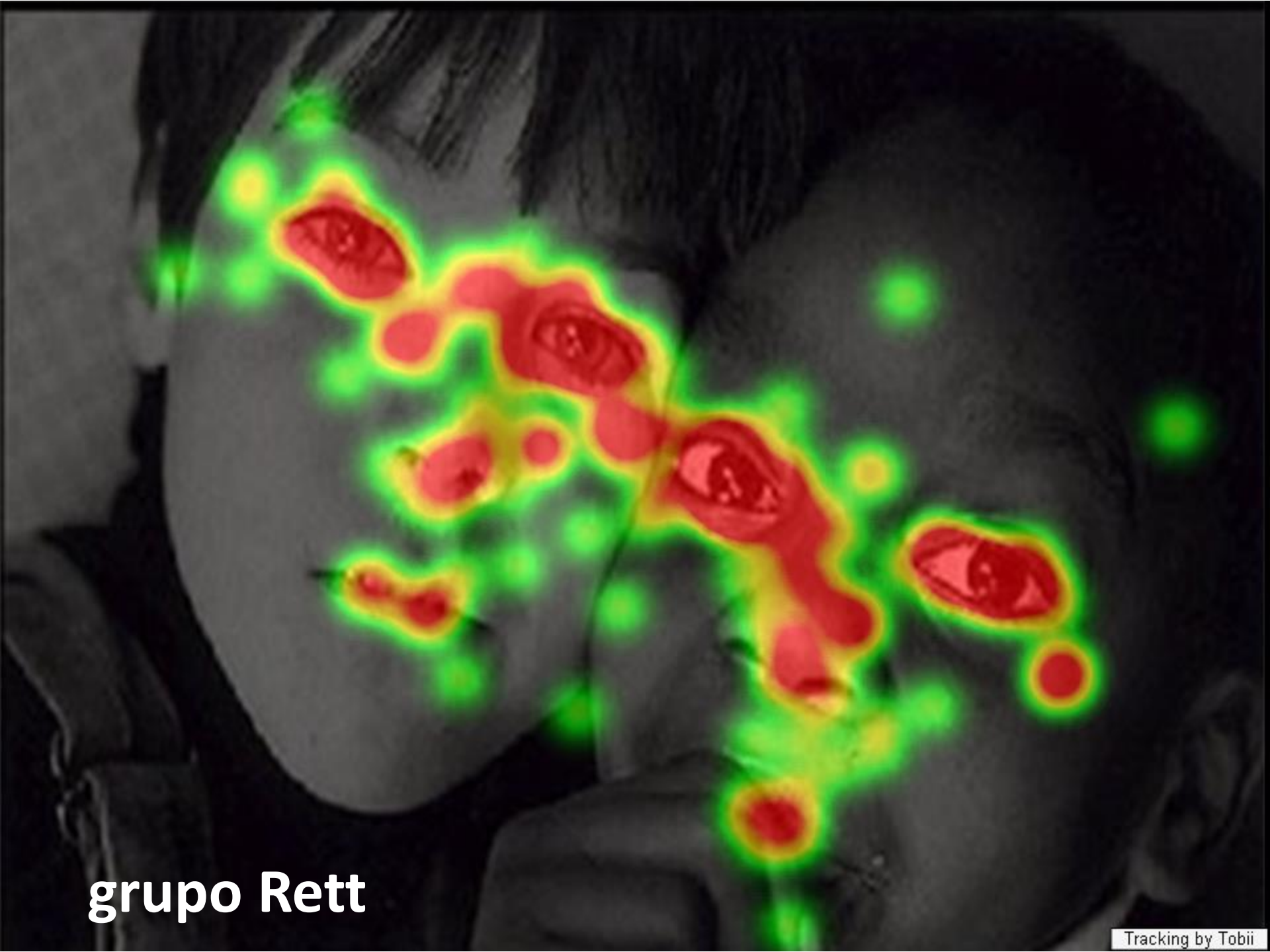


area selecionada

12
1
2
3
4
5
6
7
8
9
10
11

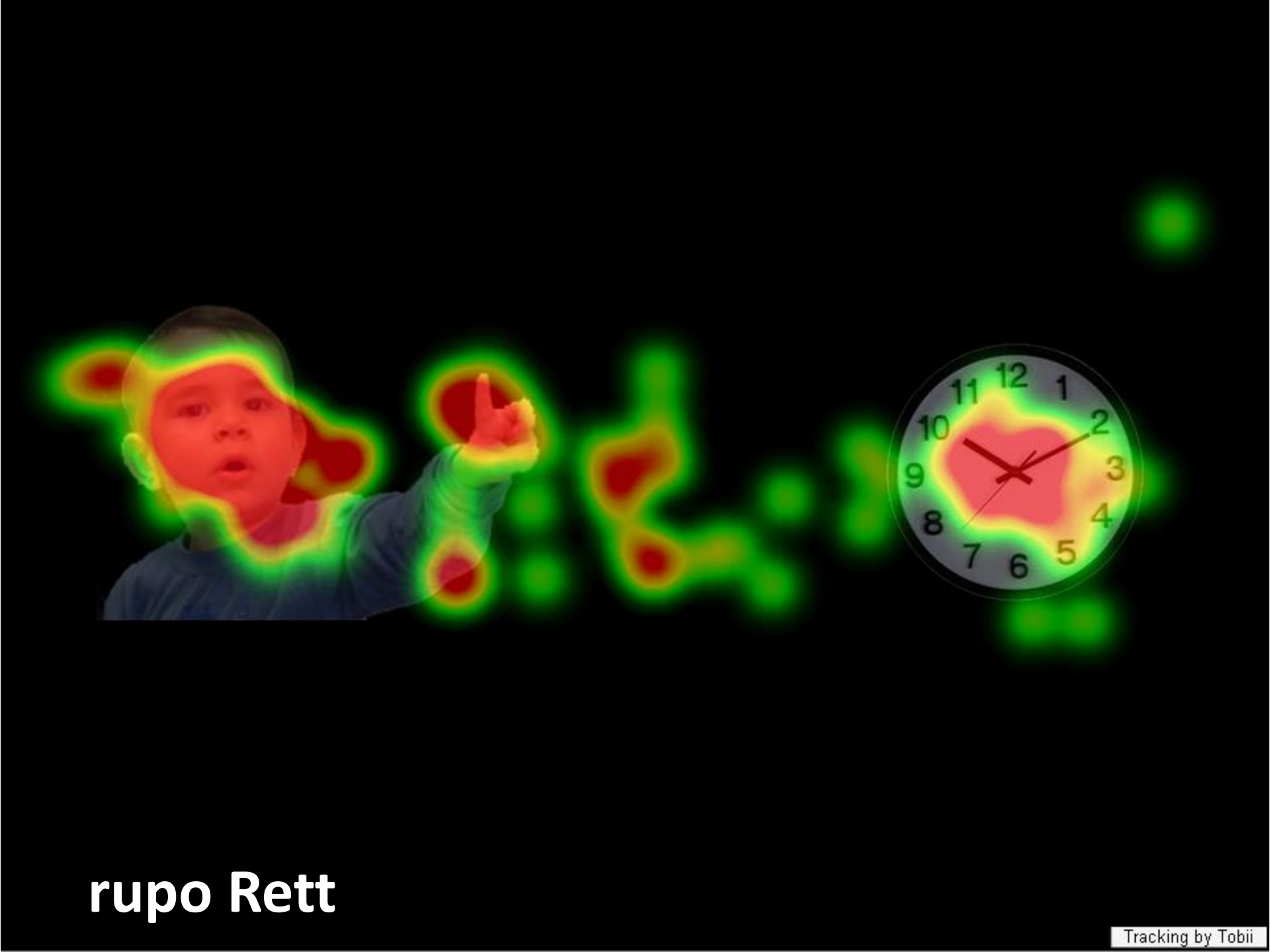
areas selecionadas




grupo Rett
Tracking by Tobii

grupo Rett

Tracking by Tobii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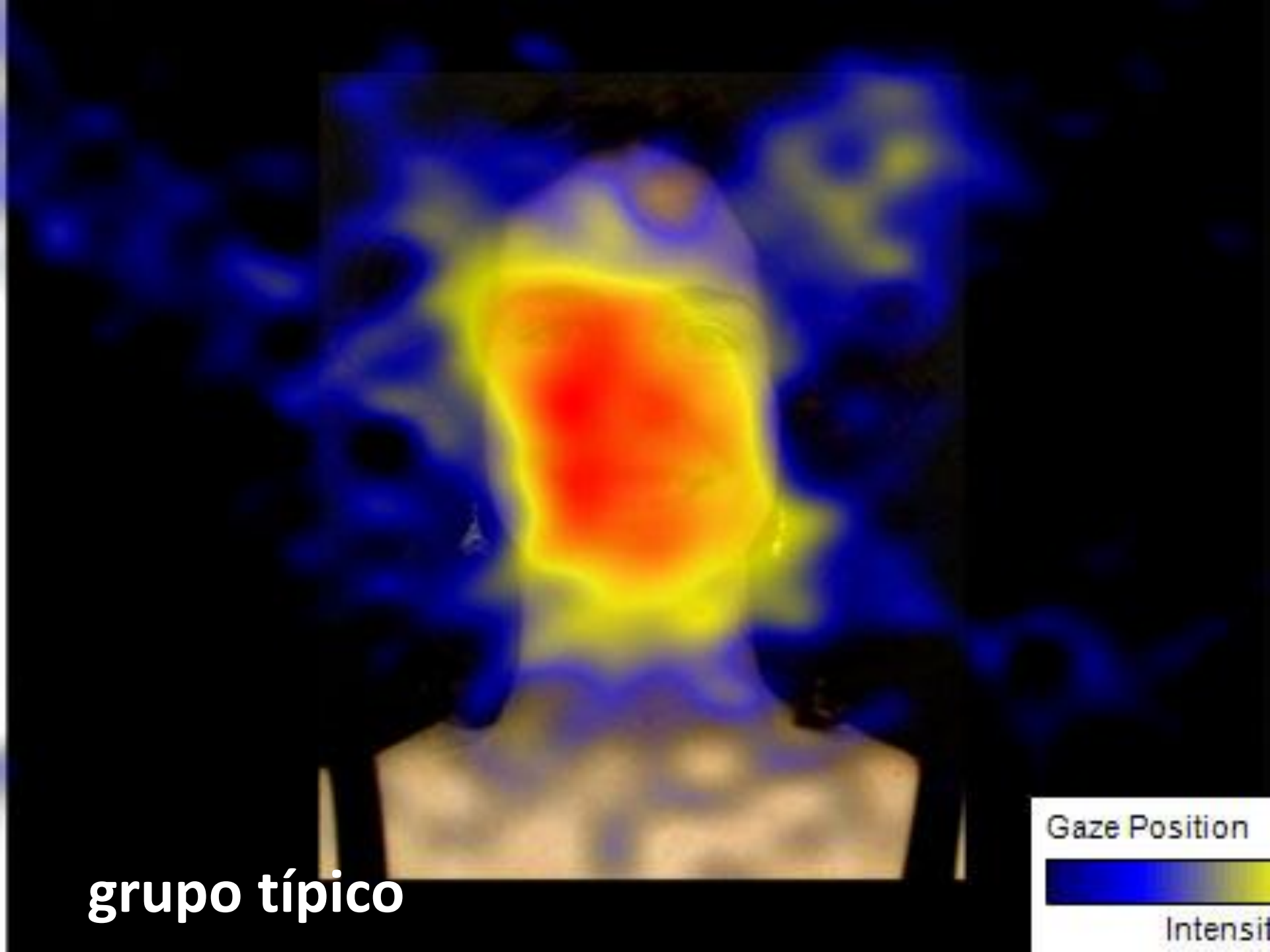

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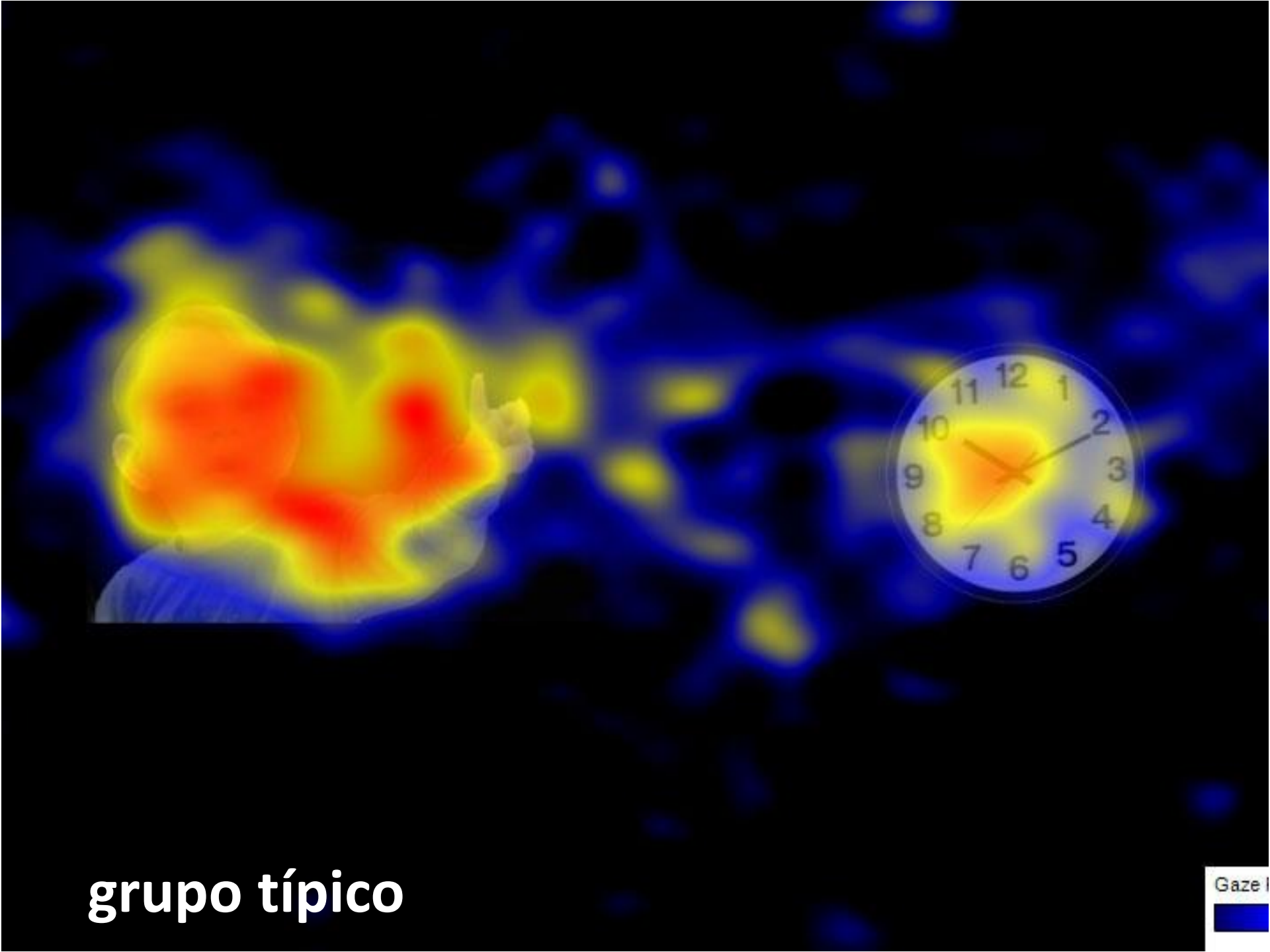

grupo típico

grupo TEA
Tracking by Tobii

grupo TEA
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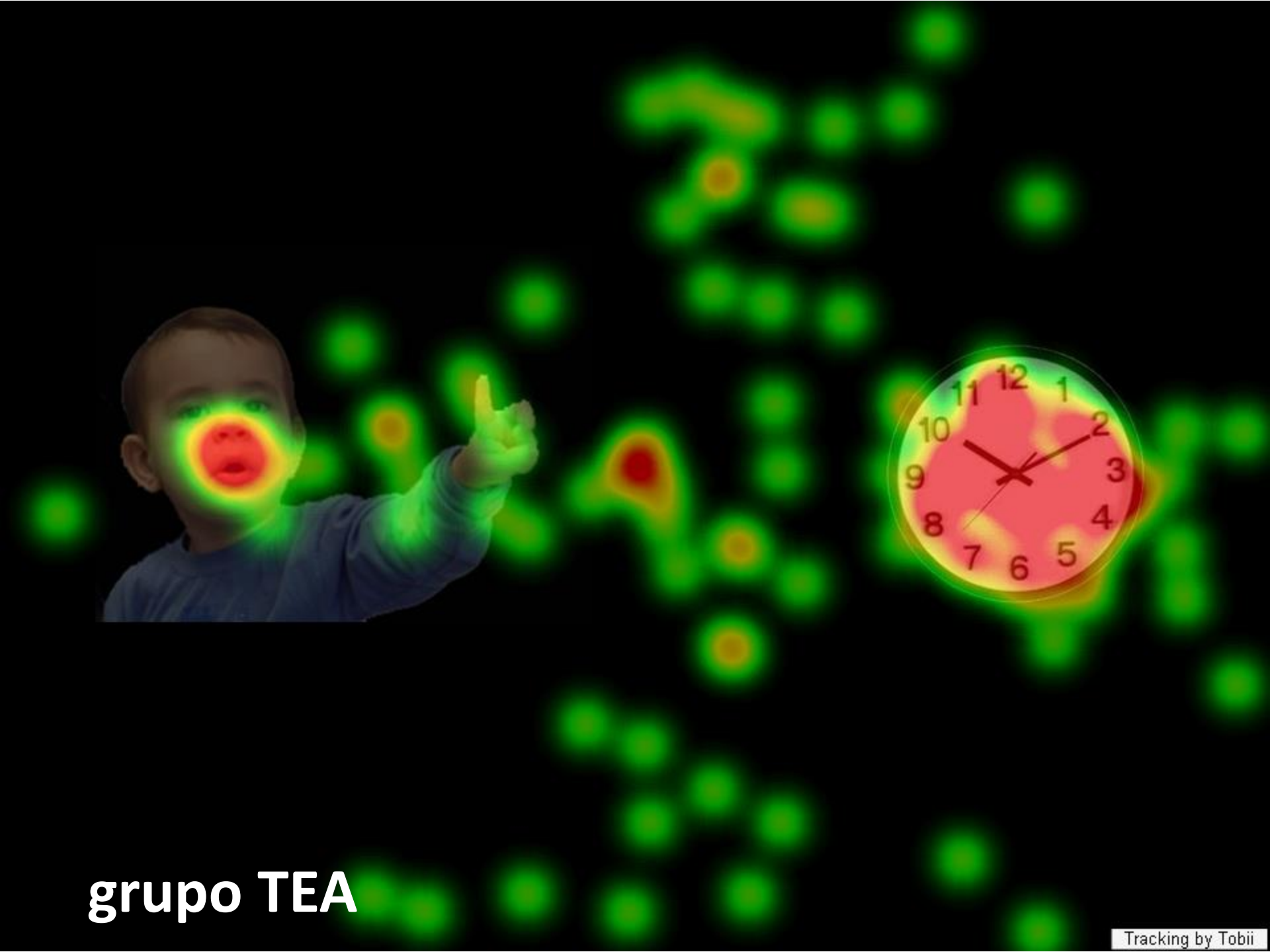
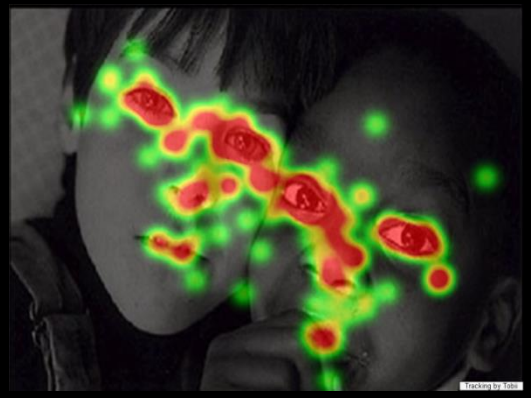
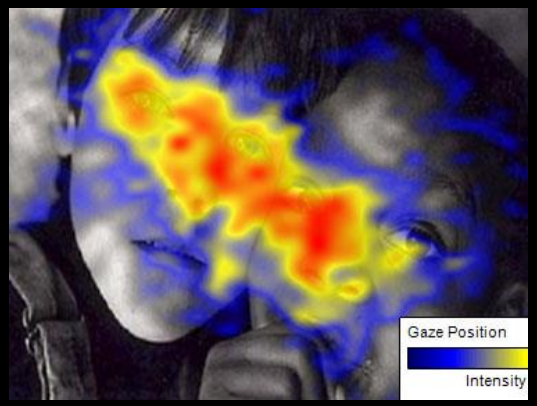
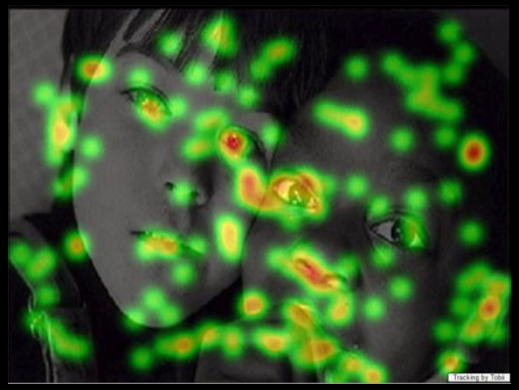
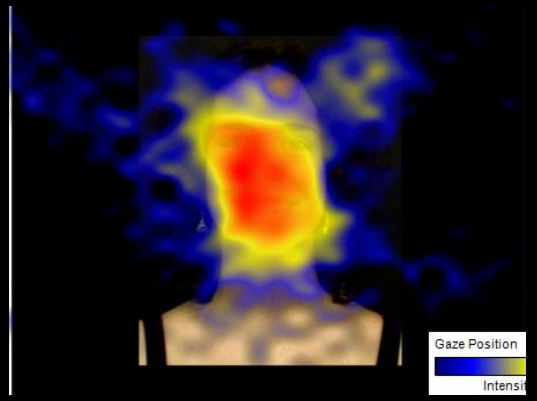
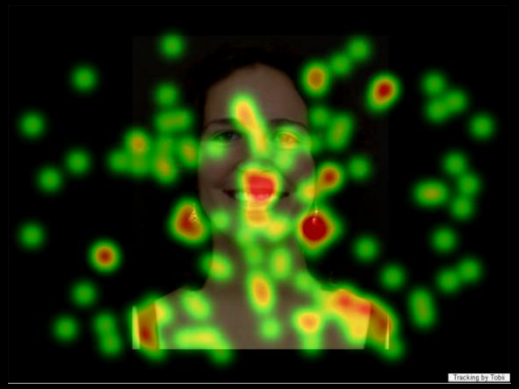
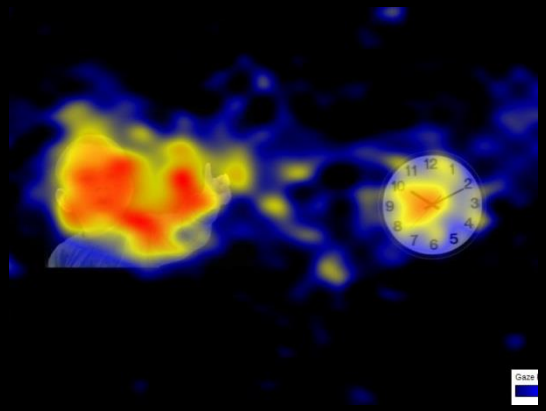
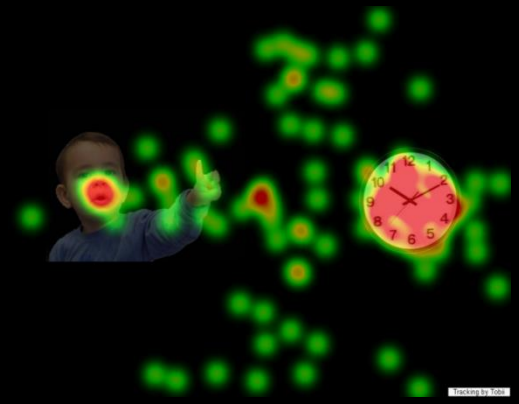

grupo TEA
Tracking by Tobii

Síndrome de Rett	Típicas	TEA
		
		
		

Preferências sociais

	Rett	Típicas	TEA		RETT	TD	0,001
P1	87,7	57,2	35,7			ASD	0,001
	9,3	31,0	23,3		DT	ASD	0,061
	Rett	Typical	ASD		RETT	TD	0,000
P2	76,6	33,7	13,5			ASD	0,001
	25,6	24,5	11,7		DT	ASD	0,017
	Rett	Typical	ASD		RETT	TD	0,002
P3a	75,8	44,5	17,8			ASD	0,000
	15,7	30,0	9,3		DT	ASD	0,008
	Rett	Typical	ASD		RETT	TD	0,134
P3b	9,2	17,9	69,5			ASD	0,000
	4,1	19,9	10,9		DT	ASD	0,000

Figura 1

Figura 2

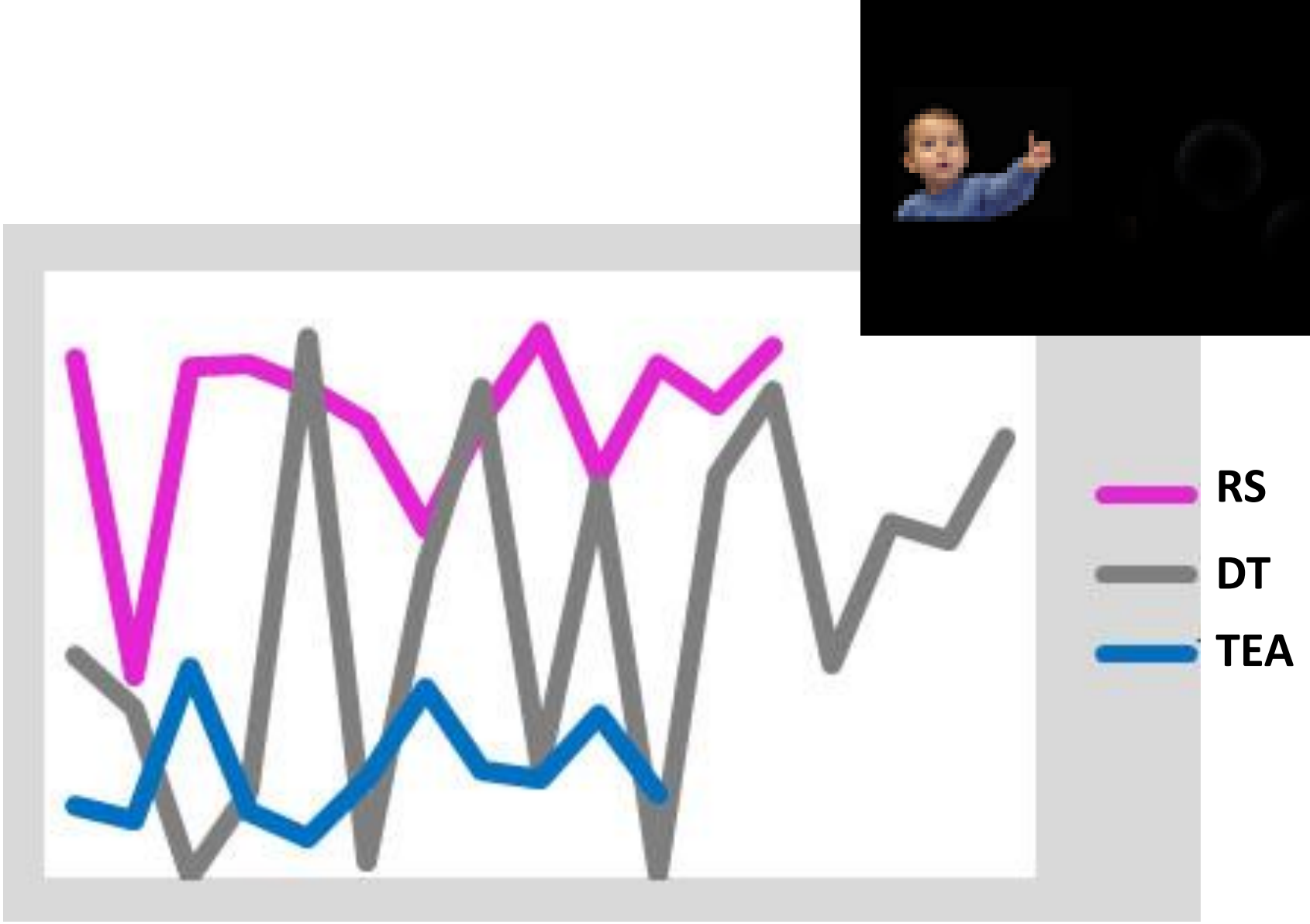


Figura 3a

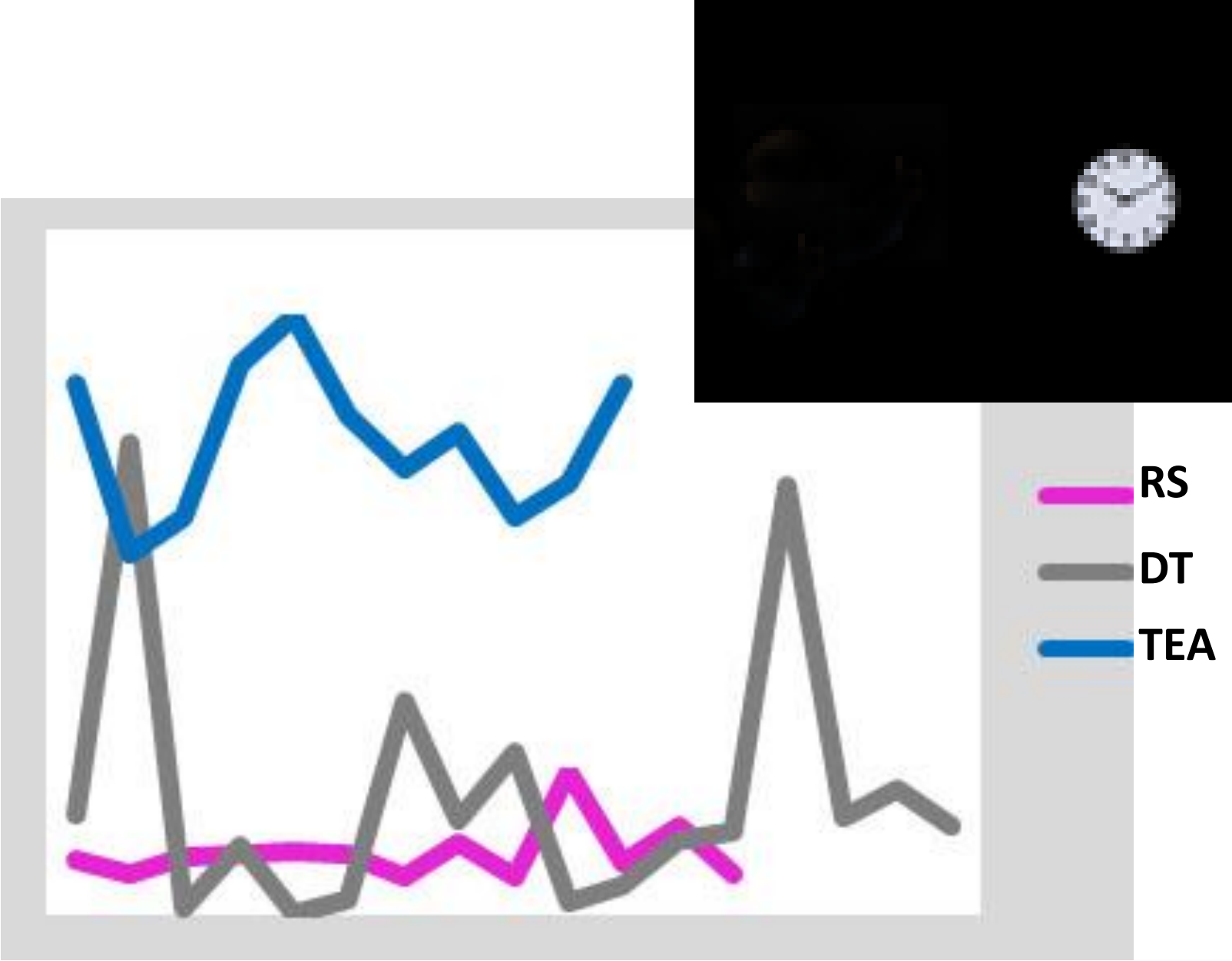
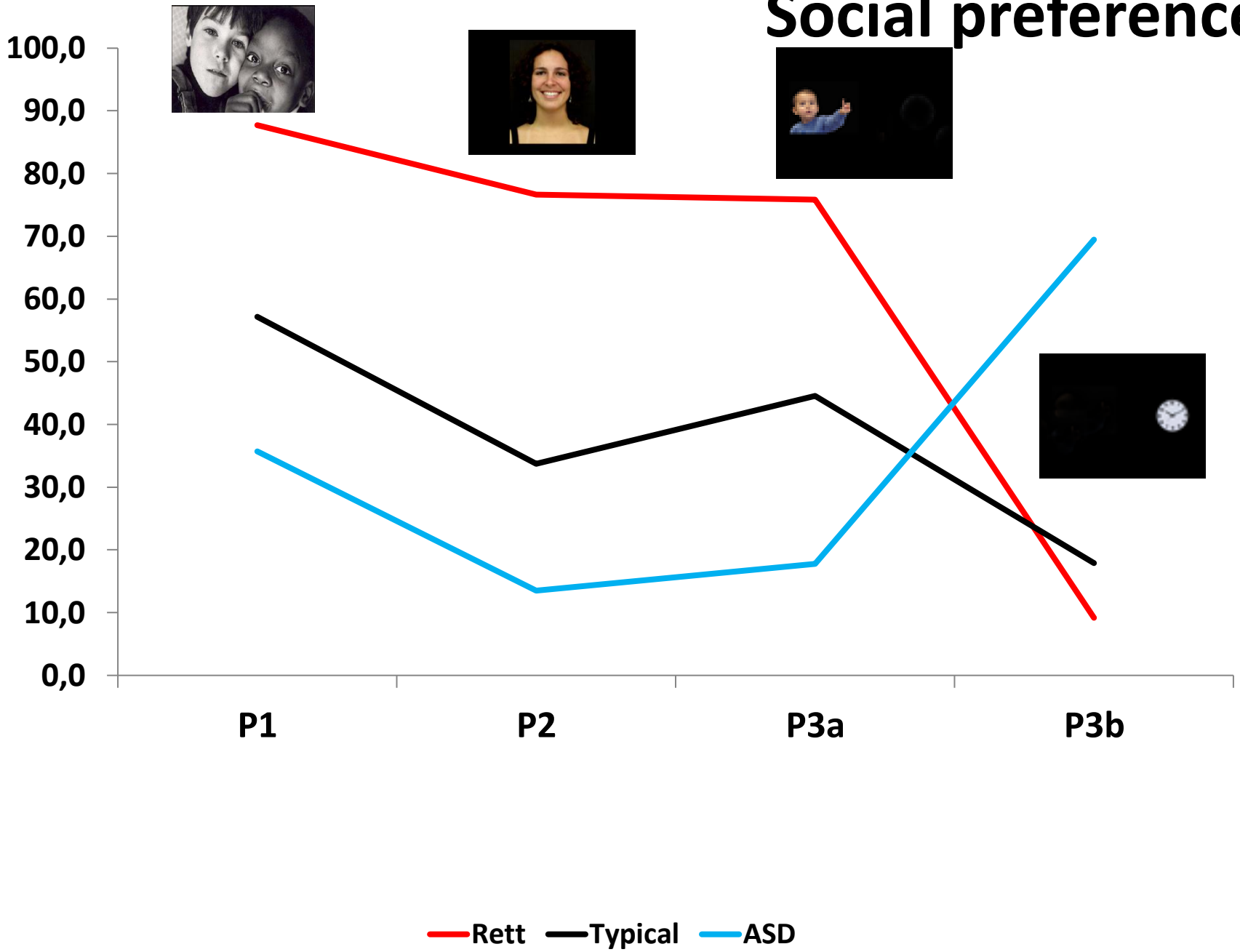


Figura 3b

Social preferences



Comments

- Pessoas com SR demonstraram padrões similares ou mesmo superiores à meninas típicas ao olhar para faces humanas.
- Demonstraram padrão inteiramente diferente quando comparadas a sujeitos com TEA
- Este pode ser considerado outro endofenótipo que difere daquele observado em pacientes com TEA.

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